ANDROGEN RECEPTOR GENE MUTATIONS DATABASE

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Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Androgen Binding: Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0312 Prostate cancer	Substitut.	5'UTR										+2 pos from transcription initiation site AR-TIS II (G>T)	Male	Normal		Crociotto et al. J Urol 158: 1599-1601, 1997
1051 Breast Cancer	Substitut.	5'UTR		105	-1101 / -1101T	105T >A						Mutation present in primary breast tumors from 2 patients	Female	Normal		Peter et al. BMC Cancer 12:132, 2012
0313 Prostate cancer	Substitut.	5'UTR		203	-913 / -913C	203C >A						+214 pos from transcription initiation site AR-TIS II	Male	Normal		Crociotto et al. J Urol 158: 1599-1601, 1997
0754 PAIS	Substitut.	1 Nterm	*	002 / 1119	4 / c.4G	p.(Glu2 Lys) / 1119G >A					high	20-50% reduction in mutant protein	Male	Ambiguous	pos	Choong et al. J Clin Invest. 98: 1423-1431, 1996
0960 CAIS	Substitut.	1 Nterm	*	002 / 1119	4 / c.4G	p.(Glu2 Lys) / 1119G >A	27	13	low	high		Mother heterozygous carrier	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1059 CAIS	Substitut.	1 Nterm		005 / 1129	14 / c.14T	p.(Leu5 *) / 1129T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0751 CAIS	Deletion	1 Nterm		007 / 1134	19 / c.19	p.(Leu7 fs) / 1134 del C						Elevated testosterone stop in codon 33 No WD development	Female	Normal	neg	Barbaro et al. Clin Endocrinol 66: 822 -826, 2007
0963 CAIS	Duplicat.	1 Nterm		015 / 1154	39 / c.39_42	p.(Pro15 fs) / 1154_1157 dupGCCG	20	9				4 nt duplication (GCCG) causing fs and stop in codon 84	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1060 CAIS	Substitut.	1 Nterm		020 / 1173	58 / c.58	p.(Arg20 *) / 1173C >T										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
1061 CAIS	Substitut.	1 Nterm		024 / 1185	70 / c.70	p.(Gln24 * / 1185C >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0624 CAIS	Deletion	1 Nterm		039 / 1230	115 / c.115_117	p.(Pro39 del) / 1230_1232 del CCC						In frame deletion of 3 nt removing proline				Jung et al. Human Genetics 114: 222, 2004
0836 CAIS	Deletion	1 Nterm		040 / 1233	118 / c.118	p.(Arg40 fs) / 1233 delA						1 nt del causing framshift & stop in codon 174	Female	Normal	pos	Decaestecker et al. Fertility & Sterility 89: 1260 e3-7, 2008
0002 CAIS	Deletion	1 Nterm		042 / 1240	125 / c.125	p.(Pro42 fs) / 1240 delC			zero			1 nt del causing frameshift & stop in Codon 174 1 aff sib	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0910 Prostate cancer	Substitut.	1 Nterm		043 / 1243	128 / c.128A	p.(Glu43 Gly) / 1243A >G						Treated with Flutamide -occurred in two cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0003 Prostate cancer	Substitut.	1 Nterm		054 / 1276	161 / c.161T	p.(Leu54 Ser) / 1276T >C						Also Phe892Leu 3789T>C mut. Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0897 CAIS	Insertion	1 Nterm		055 / 1278	163 / c.163_164	p.(Leu55 fs) / 1278_1279 insC							Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
0808 Liver cancer	Substitut.	1 Nterm		057 / 1285	170 / c.170T	p.(Leu57 Gln) / 1285T >A						Somatic mutation	Male	Normal		Yeh et al. Int J Cancer 120: 1610-1617, 2007
0005 Prostate cancer	Substitut.	1 Nterm		057 / 1285	170 / c.170T	p.(Leu57 Gln) / 1285T >A						Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0786 MAIS	Duplicat.	1 Nterm		057		p.(Leu57 dup)						Leu duplicated - male inferility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
0004 Laryngeal cancer	Deletion	1 Nterm		057								30 nt deletion Somatic mutation	Male	Normal		Urushibata et al. 10th. Int. Cong. Endocrinol Abstr. P3-706, 1996
0815 Testicular cancer	Deletion	1 Nterm		057 / 1284	169 / c.169_171	p.(Leu57 del) / 1284_1286 delCTG						3 nt deletion seminoma	Male	Normal		Garolla et al. Encdorine Related Cancer 12: 645 -655, 2005
1064 PAIS	Deletion	1 Nterm		057 / 1284	169 / c.169_171	p.(Leu57 del) / 169_171 delCTG							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012

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0411	Mental Retard.	Deletion	1 Nterm	058			8		normal	normal		3 affected sibs - CAG = 8; normal maternal uncle CAG = 23	Male	Normal	pos	Kooy et al. Am J Med Genet 85: 389-393. 1999
1062	CAIS	Substitut.	1 Nterm	058 1287	172 c.172C	p.(Gln58 *) 1287C >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0612	MAIS ?	Substitut.	1 Nterm	058 1288	173 c.173A	p.(Gln58 Leu) 1288A >T						2 out of 62 patients with male infertility	Male	Normal		Lund et al. Fertility & Sterility 79: 1647-148, 2003
0006	Kennedy Syndrome	Insertion	1 Nterm	058-080			> 40					Expansion of polyglutamine repeat	Male	Normal		LaSpada et al. Nature 352: 77-79, 1991
0007	Prostate cancer	Deletion	1 Nterm	058-080			18					Contraction of poly Gln repeats (24 to 18) Somatic mutation	Male	Normal		Schoenberg et al. Biochem Biophys Res Comm 198: 74-80 1994
0324	Prostate cancer	Deletion	1 Nterm	058-080			22					Deletion of 1polyGln repeat (23-22) Somatic mutation	Male	Normal		Watanabe et al. Jpn J Clin Oncol 27: 389 -393, 1997
0325	Prostate cancer	Insertion	1 Nterm	058-080			22					Insertion of 1polyGln repeat (21-22) in 2 diff patients.Som mut	Male	Normal		Watanabe et al. Jpn J Clin Oncol 27: 389 -393, 1997
0495	Prostate cancer	Deletion	1 Nterm	058-080			18					Contraction of poly Gln repeats (20 to 18) Somatic mutation	Male	Normal		Wallin et al. J Pathology 189: 559-653, 1999
0692	CAIS	Substitut.	1 Nterm	059 1291	176 c.176C	p.(Gln59 *) 1291C >T			zero				Female	Normal		Holterhus et al. J Mol Med (Berl) 83: 1005 -1013, 2005
0008	CAIS	Substitut.	1 Nterm *	060 1293	178 c.178C	p.(Gln60 *) 1293C >T			low	normal	high	Normal upregulation.	Female	Normal	neg	Zoppi et al. J Clin Inv 19: 1105, 1993
0671	CAIS	Substitut.	1 Nterm *	060 1293	178 c.178C	p.(Gln60 *) 1293C >T						bilateral inguinal hernia.	Female	Normal		Bouvattier et al. J Clin Endocrinol Metab 87: 29-32, 2002
1063	CAIS	Substitut.	1 Nterm	060 1293	178 c.178	p.(Gln60 *) 1293C >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0409	CAIS	Duplicat.	1 Nterm	060 1294	179 179	p.(Gln60 fs) 1294 dupA						either 1 nt insert or 2 nt del. - frameshift & stop in codon 83	Female	Normal		Zhu et al. J Clin Endocrinol Metab 84: 1590-1594, 1999
0009	Prostate cancer	Substitut.	1 Nterm	064 1303	188 c.188A	p.(Gln64 Arg) 1303A >G						Also Leu831Pro 3607T>C mut. Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0846	CAIS	Substitut.	1 Nterm	067 1311	196 c.196C	p.(Gln67 *) 1311C >T						Diag at 1Mo Bilateral gonadectomy at 18y	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
0894	CAIS	Substitut.	1 Nterm	070 1323	208 c.208C	p.(Gln70 *) 1323C >T							Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
0881	MAIS ?	Substitut.	1 Nterm	070 1324	209 c.209A	p.(Gln70 Arg) 1324A >G	20, 21, 23					22 CAGs in blood azoospermia Sertoli Cell only Syndrome	Male	Normal		Hose et al. Fertility & Sterility 92: 390e9-e11, 2009
0787	PAIS?	Substitut.	1 Nterm	073 1332	217 c.217C	p.(Gln73 *) 1332C >T						Somatic mosiac 2/3 mutant to 1/3mutant	Female	Normal		Mueller et al. Hum Genet 119: 681, 2006
0895	CAIS	Substitut.	1 Nterm	076 1341	226 c.226C	p.(Gln76 *) 1341C >T							Female	Normal	pos	Philibert et al. Fertility & Sterility 94: 472-476 2010
0961	CAIS	Substitut.	1 Nterm	076 1341	226 c.226C	p(.Gln76 *) 1341C >T		18				Truncated CAG repeat ?	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0902	CAIS	Deletion	1 Nterm	076 1342	227 c.227	p.(Gln76 del) 1342 delA							Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
0807	Liver cancer	Substitut.	1 Nterm	080 1355	240 c.240A	p.(Gln80) 1355A >G						somatic mutation? present in tumor and non-tumor tissue	Male	Normal		Yeh et al. Int J Cancer 120: 1610-1617, 2007
0962	CAIS	Insertion	1 Nterm	081 1355	240 c.240_241	p.(Glu81 fs) 1355_1356 insA	21	18				1 nt insertion causing frameshift and stop in codon 83	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0901	CAIS	Deletion	1 Nterm	082 1359	244 c.244_248	p.(Thr82 fs) 1359_1363 delACAG						5 base deletion	Female	Normal	pos	Philibert et al. Fertility & Sterility 94: 472-476 2010
0965	CAIS	Deletion	1 Nterm	084		p.(Ser83 fs)	20	18	zero	zero		10 base deletion frameshift & stop in codon 171	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010

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0882 Prostate Cancer	Deletion	1 Nterm		086 1371	256 c.256_258	p.(Gln86 del) 1371_1373 delCAG						also p.Q58L,T440P, G456S, AR23 splice var. flutamide treatmt	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0964 CAIS	Substitut.	1 Nterm		086 1371	256 c.256C	p(.Gln86 *) 1371C >T	20		low	high			Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0416 CAIS	Insertion	1 Nterm		087 1372	257 c.257_258	p.(Gln87 fs) 1372_1373 insA	25		zero			1 nt insertion causing frameshift and stop in codon 93	Female	Normal		Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0796 CAIS	Substitut.	1 Nterm		088 1377	262 c.262C	p.(Gln88 *) 1377C >T			zero				Female	Normal		Jaaskelainen et al. Hum Mutat 27: 291, 2006
0672 CAIS	Substitut.	1 Nterm		090 1383	268 c.268C	p(.Gln90 *) 1383C >T						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endocrinol Metab 87: 29-32, 2002
1065 CAIS	Substitut.	1 Nterm		090 1383	268 c.268C	p(.Gln90 *) 1383C >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0806 Liver cancer	Substitut.	1 Nterm		091 1385	270 c.270G	p.(Gln90 His) 1385G >T						Somatic mutation	Male	Normal		Yeh et al. Int J Cancer 120: 1610-1617, 2007
1066 CAIS	Substitut.	1 Nterm		091 1386	271 c.271C	p.(Gln91 *) 1386C >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
1067 CAIS	Substitut.	1 Nterm		093 1393		p.(Glu93 *) 1393C >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0529 CWR22R Prost. CA Cell line	Substitut.	1 Nterm		093 1395		p.(Glu93 Asp) 1395G	27	19				AR indep.+p.L57Q & H875Y mut. + Duplication of exon 3	Male	Normal		Chelnski et al. The Prostate 47: 66-75, 2001
0805 Liver cancer	Substitut.	1 Nterm		102 1417	302 c.302G	p.(Arg101 His) 1417G >A						somatic mutation ? present in tumor and non-tumor tissue	Male	Normal		Yeh et al. Int J Cancer 120: 1610-1617, 2007
0417 CAIS	Deletion	1 Nterm		104 1427	312 c.312	p.(Pro104 fs) 1427 delC	12	25	zero			1 nt deletion causing frameshift and stop in codon 174	Female	Normal	neg	Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0898 CAIS	Insertion	1 Nterm		109 1440	325 c.325_326	p.(Val109 fs) 1440_1441 insA							Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
0010 Prostate cancer	Substitut.	1 Nterm		114 1457	342 c.342G	p.(Gln114 His) 1457G >T						Also Trp797* 3506G>A mut. Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0418 CAIS	Substitut.	1 Nterm		115 1458	343 c.343C	p.(Gln115 *) 1458C >T	25	27					Female	Normal		Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0802 CAIS	Substitut.	1 Nterm		120 1473	358 c.358C	p.(Gln120 *) 1473C >T						1 affected sister	Female	Normal	pos	Berg et al. J Pediatr 150: 434-438, 2007
1068 CAIS	Substitut.	1 Nterm		120 1473	358 c.358C	p.(Gln120 *) 1473C >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
1068 PAIS	Substitut.	1 Nterm		120 1473	358 c.358C	p.(Gln120 Glu) 1473C >G										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0631 CAIS	Substitut.	1 Nterm		121 1477	362 c.362C	p.(Ser121 *) 1477C >A	25					gonads located in inguinal canal. Same family as 0830	Female	Normal	pos	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
0830 CAIS	Substitut.	1 Nterm		121 1477	362 c.362C	p.(Ser121 *) 1477C >A						gonads located in inguinal canal. Same family as 0631	Female	Normal	pos	Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
0911 Prostate cancer	Substitut.	1 Nterm		121 1478	363 c.363G	p.(Ser121) 1478 >C						Silent mutation. Both treated and untreated. Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0417 CAIS	Deletion	1 Nterm		127 1496	381 c.381	p.(Pro127 fs) 1496 delC	23	24	zero			1ntdeletion causing frameshift and stop at codon 174	Female	Normal		Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0011 CAIS	Deletion	1 Nterm		129 1502	387 c.387	p.(Arg129 fs) 1502 delA			zero			1 nt deletion causing frameshift & stop in codon 174	Female	Normal	neg	Batch et al. Hum Mol Genet 1: 497-503, 1992
0436 CAIS	Deletion	1 Nterm		129 1502	387 c.387	p.(Arg129 fs) 1502 delA						1 nt deletion causing frameshift & stop in codon 174	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
1058 CAIS	Substitut.	1 Nterm		131 1509	393 c.393	p.(Cys131 *) 1509C >A							Female	Normal		Cai et al. J Obstet Gynaecol 32:707-708, 2012

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0012	CAIS	Deletion		142								Frameshift & deletion of codons 142-150 Stop in codon 156	Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0837	CAIS	Substitut.		143 1542	427 c.427A	p.(Lys143 *) 1542A >T						Sertoli-Leydig tumor	Female	Normal		Jazabeck et al. Gynecol Endocrinol 23: 499 -504, 2007
0694	Prostate Cancer	Substitut.	*	144 1546	431 c.431G	p.(Gly144 Val) 1546G >T	16		normal	normal		Increased response to DHT	Male	Normal		Chen et al. The Prostate 63: 395-406, 2005
0516	CAIS	Substitut.		155 1575	460 c.460G	p.(Glu155 *) 1575G >T							Female	Normal		Copelli et al. Asian J Androl 1: 73-77, 1999
0523	CAIS	Substitut.		155 1575	460 c.460G	p.(Glu155 *) 1575G >T							Female	Normal		Gacobini et al. Hum Genet. 108. 176, 2001
0788	PAIS	Substitut.		159 1590	475 c.475G	p.(Ala159 Thr) 1590G >A										Mueller et al. Hum Genet 119: 681, 2006
0627	Prostate Cancer	Substitut.		168 1617	502 c.502G	p.(Gly168 Ser) 1617G >A						Orch + bicalutamitide treatment Gleason 10 Somatic mutation	Male	Normal		Haapala et al. Lab Invest. 81: 1647-1651, 2001
0838	CAIS	Deletion		168 1617	502 c.502	p.(Gly168 fs) 1617 delG						1 nt deletion causing frameshift & stop	Female	Normal		Jeske et al. J Pediatr Endocrinol Metab 20: 893-908, 2007
0013	CAIS	Substitut.		174 1636	521 c.521T	p.(Leu174 *) 1636T >G							Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0316	PAIS	Substitut.		174 1636	521 c.521T	p.(Leu174 *) 1636T >G			low	normal		Somatic mosaic mut. causes partial virulization	Female	Ambiguous		Holterhus et al. J Clin Endocrinol. 82: 3584-3589, 1997
0420	CAIS	Substitut.		174 1636	521 c.521T	p.(Leu174 *) 1636T >G	26	24	zero				Female	Normal	neg	Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0566	Prostate cancer	Substitut.		178 1649	534 c.534C	p.(Ser178) 1649C >T						Som mut. Estrogen treated. +p.Leu745 3350C>T silent mut	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
0014	Prostate cancer	Substitut.		182 1660	545 c.545A	p.(Lys182 Arg) 1660A >G						Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0912	Prostate cancer	Substitut.		194 1695	580 c.580C	p.(Leu194 Phe) 1695C >T						Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442
0913	Prostate cancer	Substitut.		194 1696	581 c.581T	p.(Leu194 Arg) 1696T >G						Treated with anti-androgens	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442
0319	CAIS	Substitut.		196 1702	587 c.587A	p.(Gln196 Arg) 17022A >G						Also 1 nt deletion in Codon 598 causing a stop in codon 623	Female	Normal		Komori et al. J Obstetrics & Gynocol. 23: 277-81, 1997
0551	Prostate cancer	Substitut.		200 1714	599 c.599A	p.(Glu200 Gly) 1714A >G						Treated with bicalutamide - somatic mutation	Male	Normal		Taplin et al. 37th meeting ASCO 20: Abstr, 1738 2001
0015	CAIS	Duplicat.		202 1721	606 c.606_609	p.(Glu204 fs) 1721_1724 dupATCC			zero			4 nt insertion causing frameshift	Female	Normal	neg	Batch et al. Hum Mol Genet 1: 497, 1992
0549	Prostate cancer	Substitut.		204 1727	612 c.612A	p.(Glu204 Glu) 1727A >G						Treated with bicalutamide - silent mutation- somat. mut.	Male	Normal		Taplin et al. 37th meeting ASCO 20: Abstr, 1738 2001
0395	Normal	Substitut.		207 1736	621 c.621C	p.(Ser207 Arg) 1736C >G						Homosexual individual	Male	Normal		Macke et al. Am J Human Genetics 53: 844-852, 1993
0437	CAIS	Deletion		210 1734	619 c.619	p.(Arg210 fs) 1734 delG			zero			Frameshift	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0376	MAIS	Substitut.		212 1751	636 c.636G	p.(Arg212 Arg) 1751G >A						Silent mutation	Male	Normal		Wang et al. Clinical Genetics 54: 185-192, 1998
0328	Normal	Substitut.		213 1754	639 c.639G	p.(Glu213) 1754G >A						Silent mutation - polymorphism detected in 8% popul.	Male	Normal		Batch et al. Hum Mol Genet 1: 497, 1992
0329	Normal	Substitut.		213 1754	639 c.639G	p.(Glu213) 1754G >A						Silent mut.polymorph -detected in 14% of X chromosomes	Male	Normal		Hiort et al. Eur J Pediatr 153: 317-321, 1994
0330	Normal	Substitut.		213 1754	639 c.639G	p.(Glu213) 1754G >A						Silent mutation polymorphism	Male	Normal		Lu et al. Clinical Genetics 49: 323-324. 1996

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0377 Normal	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A						Silent mutation polymorphism	Male	Normal		Wang et al. Clinical Genetics 54: 185-192, 1998
0396 Normal	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A						Silent mut.polymorph detected in 10% of X chromosomes	Male	Normal		Macke et al. Am J Human Genetics 53: 844-852, 1993
0378 MAIS	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A						Silent mutation polymorphism - 4 patients with	Male	Normal		Wang et al. Clinical Genetics 54: 185-192, 1998
0421 CAIS	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A	22	24	v low			Silent mutation - negligible level of mRNA & hAR	Female	Normal		Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0422 CAIS	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A	21	23	normal	normal		Silent mutation -	Female	Normal	neg	Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0423 PAIS	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A	23	24	v low			Silent mutation -	Male	Ambiguous		Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0424 PAIS	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A	19	24	normal	high		Silent mutation -	Male	Ambiguous	pos	Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0425 MAIS	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A	20	16	normal	high		Silent mutation -	Male	Normal		Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0705 MAIS	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A				normal			Male	Ambiguous		Deeb et al. Clinical Endocrinol 63: 56-62, 2005
0756 Prostate cancer	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006
0757 Prostate cancer	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006
0758 Prostate cancer	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006
0914 Prostate cancer	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1744G >A						Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
1019 Premature ovarian failure	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A						Present in 17.85% of patients & 15% of controls	Female	Normal		Panda et al. Gyncol Endocrinol 2010
0701 Normal	Substitut.	1 Nterm		213 1754	639 c.639G	p.(Glu213) 1754G >A						Silent mut.polymorph 65% Ivory Coast, 28% N African freqs.	Male	Normal		Estaban et al. Am J Hum Biol 17: 690-695, 2005
0379 MAIS	Substitut.	1 Nterm	*	216 1761	646 c.646G	p.(Gly216 Arg) 1761G >A	27	23	normal	normal	norm	severe oligospermia- 20% lower transactivation	Male	Normal		Wang et al. Clinical Genetics 54: 185-192, 1998
0707 PAIS	Substitut.	1 Nterm		216 1761	646 c.646G	p.(Gly216 Arg) 1761G >A				normal			Female	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
0708 PAIS	Substitut.	1 Nterm		216 1761	646 c.646G	p.(Gly216 Arg) 1761G >A				normal			Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
0709 PAIS	Substitut.	1 Nterm		216 1761	646 c.646G	p.(Gly216 Arg) 1761G >A				high			Female	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
0380 Normal	Substitut.	1 Nterm		216 1761	646 c.646G	p.(Gly216 Arg) 1761G >A							Male	Normal		Wang et al. Clinical Genetics 54: 185-192, 1998
0016 CAIS	Duplicat.	1 Nterm		217 1764	649 c.649	p.(Ala217 fs) 1764 dupG						1 nt duplication causing frameshift & stop in Codon 234	Female	Normal	neg	Hiort et al. Hum Mol Genet 3: 1163-1166 1994
0966 CAIS	Deletion	1 Nterm		221 1778	663 c.663	p.(Ser221 fs) 1778 delC	18	17	zero	zero		I nt deletion causing frameshift & stop in codon 226	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0695 Prostate cancer	Substitut.	1 Nterm	*	223 1782	667 c.667G	p.(Asp223 His) 1782G >C	16		normal	normal		Increased response to DHT	Male	Normal	neg	Chen et al. The Prostate 63: 395-406, 2005
0548 Prostate cancer	Substitut.	1 Nterm		224 1785	670 c.670A	p.(Asn224 Asp) 1785A >G						Treated with flutamide also T879A - somatic mutation	Male	Normal		Taplin et al. 37th meeting ASCO 20: Abstr, 1738 2001
0650 CAIS	Substitut.	1 Nterm		225 1790	675 c.675C	p.(Tyr225 *) 1790C >G						Wolffian remnants	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004

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0917	Prostate cancer	Substitut.	1 Nterm	229 / 1800	685 / c.685A	p.(Thr229 Ala) / 1800A >G						Both treated and untreated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0915	Prostate cancer	Indel	1 Nterm	229 / 1800	685 / c.685_686AC delins TG / 1800_1810AC	p.(Thr229 Cys)						Untreated - Occurred in two cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0916	Prostate cancer	Indel	1 Nterm	229 / 1800	685 / c.685_686AC delins TG / 1800_1801AC	p.(Thr229 Cys)						Both treated and untreated - Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0885	Prostate cancer	Indel	1 Nterm	229 / 1800	685 / c.685_686AC delins TG / 1800_1801AC	p.(Thr229 Cys)						+Gln58Leu, Ala253Val, Trp435Leu, Flutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0706	PAIS	Indel	1 Nterm *	233 / 1812	697 / c.697_698TC delins CT / 1812_1813TC	p.(Ser233 Leu)			high		*		Male	Normal		Deeb et al. Clinical Endocrinol 63: 56-62, 2005
0531	MAIS	Substitut.	1 Nterm *	235 / 1820	705 / c.715C	p.(Asn235 Lys) / 1820C >A			normal		*	Azoospermia - transactivation 46% of wt	Male	Normal		Giwercman et al. Clin Endocrinol 54: 827 -834, 2001
0967	CAIS	Insertion	1 Nterm	241 / 1835	720 / c.720_721	p.(Lys241 *) / 1835_1836 insT	21	18					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1030	MAIS	Substitut.	1 Nterm	242 / 1838	724 / c.721A	p.(Ala242 Ser / 1838G >T						Azoopsermia	Male	Normal		Goglia et al. Fert Steril 96: 1165-1169, 2011
1049	PAIS	Substitut.	1 Nterm	244 / 1844	730 / c.730T	p.Ser244 Pro / 1844T >C						External Masculisation Scale = 10	Male	Ambiguous		Hellmann et al. Arch Dis Child 97:403-409, 2012
0789	PAIS?	Substitut.	1 Nterm	248 / 1858	743 / c.743G	p.(Gly248 Val) / 1858G >T										Mueller et al. Hum Genet 119: 681, 2006
0918	Prostate Cancer	Substitut.	1 Nterm	253 / 1873	758 / c.758C	p.(Ala253 Val) / 1873C >T						Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0883	Prostate Cancer	Substitut.	1 Nterm	255 / 1878	763 / c.763G	p.(Glu255 Lys) / 1873G >A						+L446S, R485C,K610E, R787*, L798P, L874P, AR23, Flutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0350	CAIS	Substitut.	1 Nterm *	263 / 1903	788 / c.788T	p.(Leu263 Pro) / 1903T >C					*	Also Gly821Ala mut. Extra mutation causes greater thermolability	Female	Normal		Tanaka et al. Gynocol Endocrinol 12: 75-82, 1998
0845	CAIS?	Duplicat.	1 Nterm	265 / 1908	793 / c.793	p.(Gly265 fs) / 1908 dupG										Mueller. Hum Genet 123: 105, 2008
0019	CAIS	Deletion	1 Nterm	266 / 1912	797 / c.797	p.(Asp266 fs) / 1912 delA			zero			1 nt deletion causing frameshift & stop in codon 296	Female	Normal		Bruggenwirth et al. J Steroid Biochem Mol Biol 58: 569-575,1996
0017	Prostate cancer	Substitut.	1 Nterm	268 / 1918	803 / c.803T	p.(Met268 Thr) / 1918T >C						Also p.Leu575Pro mut. Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0018	Prostate cancer	Substitut.	1 Nterm	271 / 1926	811 / c.811C	p.(Pro271 Ser) / 1926C >T						Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0780	MAIS	Substitut.	1 Nterm	272 / 1929	814 / c.814C	p.(Leu272 Phe) / 1929C >T						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
0606	CAIS	Substitut.	1 Nterm	289 / 1980	865 / c.865G	p.(Glu289 *) / 1980G >T			low	normal		low expression of WT AR - Somatic mosaicism	Female	Normal		Holterhaus et al. Genome Biology 4: R37
0556	Prostate cancer	Substitut.	1 Nterm	298 / 2009	894 / c.894C	p.(Ser298 Arg) / 2009C >A						Poor differentiation of CaP. Germline mutation ?	Male	Normal		Yu et al. Sheng Wu Hua Xue 32: 459-462, 2000
0874	Prostate cancer	Substitut.	1 Nterm	298 / 2009	894 / c.894C	p.(Ser298 Arg) / 2009C >A			normal			Transcrip activity activarted by estradiol and progesterone	Male	Normal		Li et al. Clin Exp Pharmacolo Physiol 35: 1252-1257, 2008
0816	Testicular cancer	Substitut.	1 Nterm	299 / 2010	895 / c.895G	p.(Ala299 Thr) / 2010G >A						Seminoma	Male	Normal		Garolla et al. Encdorine Related Cancer 12: 645 -655, 2005
1052	CAIS	Indel	1 Nterm	312 / 2048	933 / c.933_1219 delins 77	p.(Phe312 fs) / 2048_2335 delins 77						287 deletion + 77 insertion causing frameshit and stop	Female	Normal	pos	Cong et al. Gene 500: 220-223, 2012
0550	Prostate cancer	Substitut.	1 Nterm	336 / 2121	2121 / c.1006T	p.(Ser336 Pro) / 2121T >C						Treated with flutamide somatic mutation	Male	Normal		Taplin et al. 37th meeting ASCO 20: Abstr, 1738 2001
0398	Prostate cancer	Substitut.	1 Nterm	342 / 2140	1025 / c.1025C	p.(Pro342 Leu) / 2140C >T						Somatic mutation Stage 3 tumor	Male	Normal		Castagnaro et al. Verh. Dtsch. Ges. Path. 77: 119-123, 1993

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0779	MAIS	Substitut.		342	1025	p.(Pro342 Leu)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		1 Nterm		2140	c.1025C	2140C >T										
0968	CAIS	Deletion		348	1041	p.(Tyr348 fs)	21	17				2 nt deletion causing fs & stop in Codon 500	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		1 Nterm		2156	c.1041_1042	2156_2157 delCT										
0903	CAIS	Deletion		349	1035	p.(Lys349 fs)							Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
		1 Nterm		2150	c.1035_1036	2150_2151 delinsAG										
0903	CAIS	Deletion		349	1041	p.(Gly349 fs)						22 bp deletion	Female	Normal	pos	Philibert et al. Fertility & Sterility 94: 472-476 2010
		1 Nterm		2156	c.1041_1062	2156_2177 del										
0020	CAIS	Substitut.		355	1063	p.(Glu355 *)	21	23	low			low specific binding with MB only-mRNA < 20%	Female	Normal	neg	Gottlieb et al. Hum Mutat. 14: 527-539, 1999
		1 Nterm		2178	c.1063G	2178G >T										
0781	MAIS	Substitut.		355	1063	p.(Glu355 Gln)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		1 Nterm		2178	c.1063G	2178G >C										
0919	Prostate Cancer	Substitut.		359	1072	p.(Ala358 Thr)						Bicalutamide treated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2187	c.1072G	2187G >A										
0920	Prostate Cancer	Substitut.		359	1073	p.(Ala359 Val)						Bicalutamide treated Occurred in two cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2188	c.1073C	2188C >T										
0921	Prostate cancer	Substitut.		362	1085	p.(Arg362 His)						Both treated and untreated Occurred in two cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2200	c.1085G	2200G >A										
0021	CAIS	Substitut.		373	1117	p.(Gly373 *)						Somatic instability of polyglycine tract	Female	Normal	pos	Davies et al. Clinical Endocrinology 43: 69 -77, 1995
		1 Nterm		2232	c.1117G	2232G >T										
1003	PAIS	Substitut.		380	1138	p.(Pro380 Arg)	17	18				Prenatal diagnosis	Male	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		1 Nterm		2253	c.1138C	2253C >G										
0899	CAIS	Insertion		381	1129	p.(Pro380 fs)						11 bp duplication (CCTCCGCCGCC)	Female	Normal	pos	Philibert et al. Fertility & Sterility 94: 472-476 2010
		1 Nterm		2244	c.1129_1139	2244_2254 dup										
0338	MAIS	Substitut.	*	392	1174	p.(Pro392 Ser)						Oligospermia	Male	Normal		Hiort et al. J Clin Endocrinol Metab 85: 2810-2815, 2000
		1 Nterm		2289	c.1174C	2289C >T										
0504	MAIS	Substitut.	*	392	1174	p.(Pro392 Ser)						Oligospermia	Male	Normal		Hiort et al. J Clin Endocrinol Metab 85: 2810-2815, 2000
		1 Nterm		2289	c.1174C	2289C >T										
0767	MAIS	Substitut.		392	1174	p.(Pro392 Ser)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		1 Nterm		2289	c.1174C	2289C >T										
0908	CAIS	Substitut.		392	1174	p.(Pro392 Ser)			zero				Female	Normal		Appari et al. J Molecular Med: 87: 623 -632, 2009
		1 Nterm		2289	c.1174C	2289C >T										
0950	PAIS	Substitut.		392	1174	p.(Pro392 Ser)						Micropenis only	Male	Ambiguous		Bhangoo et al. Asian J Androl 12: 561-566, 2010
		1 Nterm		2289	c.1174C	2289C >T										
1004	PAIS	Substitut.		392	1174	p.(Pro392 Ser)	22	19	normal	normal			Male	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		1 Nterm		2289	c.1174C	2289C >T										
1070	PAIS	Substitut.		392	1174	p.(Pro392 Ser)										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		1 Nterm		2289	c.1174C	2289C >T										
0817	Testicular cancer	Substitut.		392	1174	p.(Pro392 Ser)						Seminoma	Male	Normal		Garolla et al. Encdorine Related Cancer 12: 645 -655, 2005
		1 Nterm		2289	c.1174C	2289C >T										
0608	Prostate cancer	Substitut.		392	1175	p.(Pro392 Leu)			zero				Female	Normal		Holterhaus et al. Genome Biology 4: R37
		1 Nterm		2290	c.1175C	2290C >T										
0547	Prostate cancer	Substitut.		392	1175	p.(Pro392 Leu)						Flutamide treated Also Asn757Asp - somatic mutation	Male	Normal		Taplin et al. Proc Am Soc Clin Oncol 20: 2001 Abstr. 1738
		1 Nterm		2290	c.1175C	2290C >T										
0022	CAIS	Substitut.		392	1175	p.(Pro392 Arg)	20	24	zero			+ p.Glu213 1754G>A & p.Gln445Arg 2449A>G	Female	Normal	pos	Gottlieb et al. Hum Mutat. 14: 527-539, 1999
		1 Nterm		2290	c.1175C	2290C >G										
0790	CAIS	Substitut.		395	1185	p.(Tyr395 *)							Female	Normal		Mueller et al. Hum Genet 119: 680, 2006
		1 Nterm		2300	c.1185C	2300C >G										
0896	CAIS	Substitut.		395	1185	p.(Tyr395 *)							Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
		1 Nterm		2300	c.1185C	2300C >G										

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0426	CAIS	Substitut.		405	1197	p.(Gln405 *)	28	23	zero			mRNA < 20%	Female	Normal		Gottlieb et al. Hum Mutat. 14: 527-539, 1999
		1 Nterm		2312	c.1197C	2312C >T										
1035	PAIS	Substitut.	*	407	1203	p.Arg407 Ser						Stage 3 PAIS-coregulator effcts melanoma antig-A11	Male	Ambiguous		Lagarde et al. J Biol Chem 287:10905-15, 2012
		1 Nterm		2318	c.1203C	2318 >A										
0689	PAIS	Deletion	*	411	1231	p(Leu4_1Ser413 del)						Del. of 9 nt & codons 411-413. Normal twin has same deletion	Male	Ambiguous	pos	Holterhus et al. Exp Clin Endocrin Diabetes 113: 457-463, 2005
		1 Nterm		2346	c.1231_1239	2346_2354 del										
0690	PAIS	Substitut.	*	413	1238	p.(Ser413 Asn)						Patient responded to testosterone treat at 12yrs.	Male	Ambiguous		Holterhus et al. Exp Clin Endocrin Diabetes 113: 457-463, 2005
		1 Nterm		2353	c.1238G	2353G >A										
0922	Prostate cancer	Substitut.		416	1246	p.(Gly416 Ser)						Bicalutamide treated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2361	c.1246G	2361G >A										
0923	Prostate cancer	Substitut.		416	1247	p.(Gly416 Asp)						Bicalutamide treated occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2362	c.1247G	2362G >A										
0691	PAIS	Substitut.	*	434	1301	p.(Ser434 Phe)							Male	Ambiguous		Holterhus et al. Exp Clin Endocrinol diabetes 113: 457-463,
		1 Nterm		2416	c.1301C	2416C >T										
0889	Prostate Cancer	Substitut.		435	1304	p.(Trp435 Leu)						+Q86del, T229C, T440P, T498I, V509L, Q828*, Q868*, Bical treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2419	c.1304G	2419G >T										
0924	Prostate Cancer	Substitut.		435	1304	p.(Trp435 Leu)							Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2419	c.1304G	2419G >T										
0925	Prostate Cancer	Substitut.		435	1305	p.(Trp435 Cys)						Both treated and untreated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2420	c.1305G	2420G >C										
0926	Prostate Cancer	Substitut.		440	1318	p.(Thr440 Pro)						Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2433	c.1318A	2433A >C										
0890	Prostate Cancer	Substitut.		440	1319	p.(Thr440 Ile)						+p.Gln58Leu, Gln86del, Gly457Asp	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2434	c.1319C	2434C >T										
1024	CAIS	Substitut.		442	1324	p.(Glu442 *)							Female	Normal		Luo FW et al. Zhongua Yi Xue. 28: 176-179, 2011
		1 Nterm		2439	c.1324G	2439G >T										
1027	CAIS	Substitut.		442	1324	p.(Glu442 *)							Female	Normal	neg	Sun et al. Eur J Obstet Gynecol. 153: 173-175, 2010
		1 Nterm		2439	c.1324G	2439G >T										
0759	Prostate cancer	Substitut.		451	1352	p.(Gly451 Asp)						patient lower Gleason score than patient -wt AR- som mutation				Sanchez et al. BJU Int 98: 1320-1325, 2006
		1 Nterm		2467	c.1352G	2467G >A										
0760	Prostate cancer	Substitut.		453	1358	p.(Gly453 Asp)						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006
		1 Nterm		2473	c.1358G	2473G >A										
0969	CAIS	Substitut.		455	1363	p.(Gly455 Ser)	22	17				Also p.Tyr571Cys 2830A>G. Sister & 2 aunts affected	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		1 Nterm		2478	c.1363G	2478G >A										
0888	Prostate Cancer	Substitut.		456	1366	p.(Gly456 Ser)						+p.Gln58Leu, Gln86del, Ala253Val, Bicalutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2481	c.1366G	2481G >A										
0927	Prostate Cancer	Substitut.		456	1366	p.(Gly456 Ser)						Treated with anti-androgens. Occurred in more than 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2481	c.1366G	2481G >A										
0884	Prostate Cancer	Substitut.		457	1370	p.(Gly457 Asp)						+Gln86del, Val716Met, AR23 splice variant Flutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2485	c.1370G	2485G >A										
0928	Prostate Cancer	Substitut.		457	1370	p.(Gly457 Asp)						Both treated and untreated. Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2485	c.1370G	2485G >A										
0438	CAIS	Deletion		463	1389	p.(Gly463 fs)			zero			1 nt deletion causing frameshift	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		1 Nterm		2504	c.1389	2504 delC										
0410	CAIS	Deletion		474	1421	p.(Glu474 fs)	24	22				2nt del causing frameshift & stop in cod 500- mRNA 50%	Female	Normal		Thiele et al. J Clin Endocrinol Metab 84: 1751-1753. 1999
		1 Nterm		2536	c.1421_1422	2536_2537 delAG										
0427	CAIS	Deletion		474	1421	p.(Glu474 fs)	26	26	zero			2nt del causing frameshift & stop in cod 500 -mRNA 50%	Female	Normal		Gottlieb et al. Hum Mutat. 14: 527-539, 1999
		1 Nterm		2536	c.1421_1422	2536_2537 delAG										
0693	CAIS	Deletion		474	1421	p.(Glu474 fs)	21	28	zero			2nt del causing frameshift & stop in cod 500- mRNA 50%	Female	Normal		Holterhus et al. J Mol Med (Berl) 83: 1005 -1013, 2005
		1 Nterm		2536	c.1421_1422	2536_2537 delAG										

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0782	MAIS	Substitut.	*	475	1424	p.(Ala475 Val)						male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		1 Nterm		2539	c.1424C	2539C >T										
0821	MAIS	Substitut.	*	475	1424	p.(Ala475 Val)	14	17				male infertility, low T severe hypospematogenesis	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
		1 Nterm		2539	c.1424C	2539C >T										
0822	MAIS	Substitut.	*	475	1424	p.(Ala475 Val)	24	17				male infertility, normal T	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
		1 Nterm		2539	c.1424C	2539C >T										
0823	MAIS	Substitut.	*	475	1424	p.(Ala475 Val)	19	17				male infertility, normal T, left cryptorchidism	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
		1 Nterm		2539	c.1424C	2539C >T										
0024	CAIS	Substitut.		481	1443	p.(Tyr481 *)	15	15	zero			Normal 110KD AR produced at 25% of normal level	Female	Normal		Gottlieb et al. Hum Mutat. 14: 527-539, 1999
		1 Nterm		2558	c.1443C	2558C >A										
0651	CAIS	Substitut.		481	1443	p.(Tyr481 *)						Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		1 Nterm		2558	c.1443C	2558C >A										
0804	Liver cancer	Substitut.		481	1443	p.(Tyr481)						Somatic mutation. Silent mutation	Female	Normal		Yeh et al. Int J Cancer 120: 1610-1617, 2007
		1 Nterm		2558	c.1443C	2558C >T										
0929	Prostate cancer	Substitut.		485		p.(Arg485 Cys)						Both treated and untreated. Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm														
0930	Prostate cancer	Substitut.		485		p.(Arg485 Thr)						Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm														
0546	CAIS	Deletion		488		p.(Gln488 *)						3 affected siblings Variable phenotype	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
		1 Nterm														
0439	CAIS	Deletion		489		p.(Gly489 fs)			low			5 nt deletion causing frameshift	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		1 Nterm														
0440	CAIS	Substitut.		492	1474	p.(Gly492 Ser)			low	low			Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		1 Nterm		2589	c.1474G	2589G >A										
0891	Prostate Cancer	Substitut.		498	1493	p.(Thr498 Ile)						Also p.Arg485Cys Bicalutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2608	c.1493C	2608C >T										
0931	Prostate Cancer	Substitut.		498	1493	p.(Thr498 Ile)						Both treated and not treated Occured in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2608	c.1493C	2608C >T										
0932	Prostate Cancer	Substitut.		499	1495	p.(Ala499 Thr)						Flutamide treated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2610	c.1495G	2610G >A										
0933	Prostate Cancer	Substitut.		499	1496	p.(Ala499 Val)						Flutamide treated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2611	c.1496C	2611C >T										
0936	Prostate Cancer	Substitut.		500	1500	p.(Pro500)						Treated with anti-androgens. Occurred in more than 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2615												
0025	CAIS	Substitut.		503	1508	p.(Trp503 *)							Female	Normal	pos	Bruggenwirth et al. J Steroid Biochem Mol Biol 58: 569-575, 1996
		1 Nterm		2623	c.1508G	2623G >A										
1071	CAIS	Substitut.		503	1508	p.(Trp503 *)							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		1 Nterm		2623	c.1508G	2623G >A										
0905	CAIS	Indel		503	1507	p.(Trp503 fs)						Indel between c.1507_1512, 22 nt insertion	Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
		1 Nterm		2622	c.1507_1512	2622_2629 delin										
0783	MAIS	Substitut.		507	1510	p.(Gly507 Asp)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		1 Nterm		2625	c.1510G	2635G >A										
0886	Prostate Cancer	Substitut.		509	1525	p.(Val509 Leu)						Bicalutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2640	c.1525G	2640G >C										
0934	Prostate Cancer	Substitut.		509	1525	p.(Val509 Leu)						Flutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2640	c.1525G	2640G >C										
0935	Prostate Cancer	Substitut.		509	1526	p.(Val509 Gly)						Treated with anti-androgens. Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		1 Nterm		2641	c.1526T	2641T >G										
0339	MAIS	Substitut.	*	512	1536	p.(Val512 Val)						Oligospermia caused by silent mutation	Male	Normal		Hiort et al. 80th US Endo Soc Meeting, Abstr P2-38, 1998
		1 Nterm		2651	c.1536G	2651G >A										

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0876	CAIS	Substitut.		514	1542	p.(Tyr514 *)							Female	Normal	pos	Radpour et al. J of Andrology 30: 230-232, 2009
		1 Nterm		2657	c.1542T	2657T >A										
0567	Prostate cancer	Substitut.		515	1543	p.(Pro515 Ser)						Somatic mut. Orchiectomy + EMP Horm-refractory CaP	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
		1 Nterm		2658	c.1543C	2658C >T										
0686	Normal	Substitut.		517		p.(Pro517 Ser)						Patient suffered from phobia		Normal		Yan et al. Psychiatric Genet 14: 57-60, 2004
		1 Nterm														
0568	Prostate cancer	Substitut.		525	1563	p.(Gly525 Ser)						Somatic mut. Orchi + EMP treat. Also Trp527* mutation	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
		1 Nterm		2678	c.1563G	2678G >A										
0569	Prostate cancer	Substitut.		525	1574	p.(Gly525 Asp)						Somatic mut. Orchiectomy + EMP Horm-refractory CaP	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
		1 Nterm		2689	c.1574G	2689G >A										
0026	Prostate cancer	Substitut.		529	1586	p.(Asp529 Gly)						Somatic mutation	Male	Normal		Tilley et al. Chemical Cancer Res. 2: 277-285, 1996
		1 Nterm		2701	c.1586A	2701A >G										
900	CAIS	Insertion		531	1592	p.(Tyr531 fs)						GCTA insertion	Female	Normal		Philibert et al. Fertility & Sterility 94: 472-476 2010
		1 Nterm		2707	c.1592_1593	2707_2708 insGCTA										
0570	Prostate cancer	Substitut.		534	1600	p.(Pro534 Ser)						Somatic mut. Orchiectomy + EMP horm-refractory CaP	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
		1 Nterm		2715	c.1600C	2715C >T										
0027	CAIS	Substitut.		535	1605	p.(Tyr535 *)			zero				Female	Normal	neg	McPhaul et al. FASEB J 5: 2910-15, 1991
		1 Nterm		2720	c.1605C	2720C >G										
1072	CAIS	Substitut.		535	1605	p.(Tyr535 *)			zero				Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		1 Nterm		2720	c.1605C	2720C >G										
1106	CAIS	Deletion											Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		1 Nterm														
0028	CAIS and mental retardation	Deletion							zero			Termini not yet defined	Female	Normal	neg	Trifiro et al. Mol Cell Endocrinol 75: 37-47, 1991
		1-8														
0029	CAIS	Deletion							zero				Female	Normal	pos	Quigley et al. J Clin Endocrinol Metab 74: 927-933, 1992
		1-8														
0030	CAIS	Deletion							zero				Female	Normal	pos	Hiort et al. Am J Med Genet. 63: 218-22, 1996
		1-8														
0435	CAIS	Deletion							zero				Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		1-8														
0023	CAIS	Duplicat.										Duplication of 8nt # 2787-2094 frameshift & stop in Codon 564	Female	Normal		Lumbroso et al. 10th Int Cong of Endocrinol, Abstr P1-182, 1996
		2														
1109	CAIS	Deletion										Also deletion of surrounding introns - total 6-7000 bp	Female	Normal	pos	Li et al., Genetics & Mol Res 10: 1022 -1031, 2011
		2 DBD														
0031	CAIS	Deletion											Female	Normal		Quigley et al. J Cell Biochem Suppl 16 Abstr. L323, 1992
		2														
0579	CAIS	Deletion										No immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
		2														
0580	CAIS	Deletion										No immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
		2														
0441	CAIS	Duplicat.										Duplication of exon 2	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		2														
0652	CAIS	Duplicat.										Duplication of exon 2 sister of 653	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		2														
0653	CAIS	Duplicat.										Duplication of exon 2 sister of 652	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		2														
0970	CAIS	Deletion				p.(His546 fs)	23	17				17bp del. at 2/3 splice site resulting in fs & stop in Codon 547	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		2														
0032	PAIS	Substitut.		548	1644	p.(Leu548 Phe)			low	high		Also has p. Trp742Cys 3341G>C mutation	Male	Ambiguous	pos	Melo et al. J Clin Endocrinol Metab 88: 3241-3250, 2003
		2		2759	c.1644G	2759G >T										

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0673	PAIS	Substitut.	2	548	1644	p.(Leu548 Phe)								Ambiguous		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
				2759	c.1644G	2759G >T										
0768	MAIS	Substitut.	2	548	1644	p.(Leu548 Phe)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
				2759	c.1644G	2759G >T										
0357	Prostate cancer	Deletion	2	548	1644	p.(Leu547 fs)						Frameshift - somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
				2759	c.1644	2759 delG										
0824	MAIS	Substitut.	2 *	548	1644	p.(Leu548 Phe)	19	17				Azoospermia, normal T, left varicocele, Sertoli only syndrom	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
				2759	c.1644G	2759G >T										
0825	MAIS	Substitut.	2 *	548	1644	p.(Leu548 Phe)	19	18				Male infertility, low T	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
				2759	c.1644G	2759G >T										
0033	MAIS	Substitut.	2	549	1645	p.(Pro549 Ser)						Distal hypospadia, variable penetrance in family members	Male	Near-normal male	pos	Sutherland et al. J of Urology 156: 828-831, 1996
				2760	c.1645C	2760C >T										
0358	Prostate cancer	Deletion	2	555	1665	p.(Pro555 fs)						Frameshift - somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
				2780	c.1665	2780 delA										
0359	Prostate cancer	Deletion	2	555	1665	p.(Pro555 fs)						Frameshift - somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
				2780	c.1665	2780 delA										
0952	Prostate cancer	Substitut.	2 DBD	559	1675	p.(Thr559 Ser)						Germline mutation early onset disease in African-American	Male	Normal	pos	Hu et al. Asian J Androl 12: 336-343, 2010
				2790	c.1675A	2790A >T										
0034	CAIS	Substitut.	2 DBD *	560	1679	p.(Cys560 Tyr)			normal	normal			Female	Normal	neg	Zoppi et al. Mol Endocrinol 6: 409, 1992
				2794	c.1679G	2794G >A										
0797	CAIS	Substitut.	2 DBD	563	1688	p.(Cys563 Tyr)							Female	Normal		Jaaskelainen et al. Hum Mutat 27: 291, 2006
				2803	c.1688G	2803G >A										
0840	CAIS	Duplicat.	2 DBD	563	1686	p.(563Cys dup)						Insertion of CTG	Female	Normal		Jeske et al. J Pediatr Endocrinol Metab 20: 893-908, 2007
				2801	c.1686_1688	2801_2803 dupCTG										
1036	PAIS	Substitut.	2 DBD	565	1693	p.(Asp565 Asn						External Masculisation Score = 5,	Male	Ambiguous	pos	Hellmann et al. Arch Dis Child 97:403-409, 2012
				2808	c.1693G	2808G >A										
1037	PAIS	Substitut.	2 DBD	565	1693	p.(Asp565 Asn						External Masculisation Score = 5	Male	Ambiguous	pos	Hellmann et al. Arch Dis Child 97:403-409, 2012
				2808	c.1693G	2808G >A										
0037	PAIS	Substitut.	2 DBD	569	1706	p.(Gly569 Val)			normal	normal						Chang et al. 73rd US Endo Soc Meeting, Abstr 28, 1991
				2821	c.1706G	2821G >T										
0036	PAIS	Substitut.	2 DBD	569	1706	p.(Gly569 Val)			normal			Severe hypospadia	Male	Ambiguous		Allera et al: J Clin Endocrinol Metab 80: 2697-2699, 1995
				2821	c.1706G	2821G >T										
0035	PAIS	Substitut.	2 DBD	569	1705	p.(Gly569 Trp)			normal	normal			Female	Normal		Lobaccaro et al. Clin Endocrinol, 40: 297, 1994
				2820	c.1705G	2820G >T										
0674	PAIS	Substitut.	2 DBD	569	1705	p.(Gly569 Trp)										Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
				2820	c.1705G	2820G >T										
0971	CAIS	Substitut.	2 DBD	569	1706	p.(Gly569 Glu)	26	18					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
				2821	c.1706G	2821G >A										
0971	CAIS	Substitut.	2 DBD	569	1706	p.(Gly569 Glu)							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
				2821	c.1706G	2821G >A										
0545	PAIS	Substitut.	2 DBD	572	1714	p.(Tyr572 His)	21					sibling of 0747	Male	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
				2829	c.1714T	2829T >C										
0558	PAIS	Substitut.	2 DBD	572	1714	p.(Tyr572 His)						DHT therapy effective	Male	Ambiguous		Foresta et al. Am J Med Genet 107: 259-260, 2002
				2829	c.1714T	2829T >C										
0747	PAIS	Substitut.	2 DBD	572	1714	p.(Tyr572 His)						sibling of 0545	Male	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
				2829	c.1714T	2829T >C										
0748	PAIS	Substitut.	2 DBD	572	1714	p.(Tyr572 His)			normal	low		nephew of 0545 & 0747	Male	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
				2829	c.1714T	2829T >C										
0769	MAIS	Substitut.	2 DBD	572	1714	p.(Tyr572 His)						male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
				2829	c.1714T	2829T >C										

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0826	PAIS?	Substitut.	*	572	1714	p.(Tyr572 His)	19	19				male infertility	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
		2 DBD		2829	c.1714T	2829T >C										
0032	CAIS	Substitut.	*	572	1715	p.(Tyr572 Cys)							Female	Normal		Komori et al. Arch Gynecol & Obstetrics 261: 95-100, 1998
		2 DBD		2830	c.1715A	2830A >G										
0972	CAIS	Substitut.		574	1720	p.(Ala574 Pro)	24	18				1 affected sister	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		2 DBD		2835	c.1720G	2835G >C										
0038	CAIS	Substitut.		574	1721	p.(Ala574 Asp)			normal			Defective DNA binding & transactivation	Female	Normal	neg	Bruggenwirth et al. J Steroid Biochem Mol Biol 58: 569-575, 1996
		2 DBD		2836	c.1721C	2836C >A										
0489	Prostate Cancer	Substitut.		576	1726	p.(Thr576 Ala)						Somatic mutation	Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000
		2 DBD		2841	c.1726A	2841A >G										
0810	Prostate Cancer	Substitut.	*	576	1726	p.(Thr576 Ala)						Som mut. also T878A binds to non-AR-specific motifs	Male	Normal		Monge et al.Cell Mol Life Sci 63: 487-497, 2006
		2 DBD		2841	c.1726A	2841A >G										
0040	CAIS	Substitut.		577	1730	p.(Cys577 Phe)			normal	normal			Female	Normal		Chang et al. 73rd US Endo Soc Meeting, Abstr 28, 1991
		2 DBD		2845	c.1730G	2845G >T										
0407	CAIS	Substitut.		577	1730	p.(Cys577 Phe)						Lack of DNA binding -19 members of same family 3 testis tumors	Female	Normal	pos	Hooper et al. Cinical Genetics 65: 183-190, 2004
		2 DBD		2845	c.1730G	2845G >T										
0039	CAIS	Substitut.	*	577	1729	p.(Cys577 Arg)			normal	normal			Female	Normal	pos	Zoppi et al. Mol Endocrinol 6: 409, 1992
		2 DBD		2844	c.1729T	2844T >C										
0554	PAIS	Substitut.	*	578	1732	p.(Gly578 Arg)			normal	normal	high	Alters affinity & selectivity of AR-ARE interactions				Nguyen et al. Mol Endocrinol 15: 1790 -1802, 2001
		2 DBD		2847	c.1732G	2847G >A										
0509	PAIS	Substitut.	*	579	1736	p.(Ser579 Thr)			normal			partial tranactivation in COS cells	Male	Ambiguous		Giwercman et al. Hormone Research 53: 83-88, 2000
		2 DBD		2851	c.1736G	2851G >C										
1023	CAIS	Substitut.		579	1736	p.(Ser579 Asp)							Female	Normal	pos	Xiao et al. Chinese Med J 123: 1473-1476, 2010
		2 DBD		2851	c.1736G	2851G >A										
0043	CAIS	Deletion		580	1740	p.(Cys580 fs)			zero			Single nt deletion causing frameshift & stop in codon 620	Female	Normal		Imai et al. Annals Clin Biochem 32: 482-486, 1995
		2 DBD		2855	c.1740	2855 delC										
0041	CAIS	Substitut.		580	1739	p.(Cys580 Tyr)							Female	Normal		Sultan et al. J Steroid Biochem Mol Biol: 46: 519-30, 1993
		2 DBD		2854	c.1739G	2854G >A										
0042	CAIS	Substitut.	*	580	1739	p.(Cys580 Phe)			normal	normal		Reduced transcription & DNA binding	Female	Normal	pos	Imasaki et al. Mol & Cell Endocrinol 120: 15 -24, 1996
		2 DBD		2854	c.1739G	2854G >T										
0973	CAIS	Substitut.	*	580	1740	p.(Cys580 Trp)	23	17	low	low		Mother & 1 aunt heterozygous carriers, 1 aunt affected	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		2 DBD		2855	c.1740C	2855C >G										
0487	Prostate Cancer	Substitut.		581	1742	p.(Lys581 Arg)						Somatic mutation	Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000
		2 DBD		2857	c.1742A	2857A >G										
0843	PAIS	Substitut.		581	1742	p.(Lys581 Arg)						Attempted sex reassignment at 31 did not succeed	Female	Ambiguous		Katsumata et al. Endocr J 55: 225-228, 2008
		2 DBD		2857	c.1742A	2857A >G										
0044	CAIS	Substitut.	*	582	1744	p.(Val582 Phe)			normal	normal			Female	Normal		Lumbroso et al. Fertil Steril, 60: 814, 1993
		2 DBD		2859	c.1744G	2859G >T										
0675	CAIS	Substitut.	*	582	1744	p.(Val582 Phe)						bilateral inguinal hernia	Female	Normal		Bouvattier et al. J Clin Endorinol & Metab 87: 29-32, 2002
		2 DBD		2859	c.1744G	2859G >T										
0676	CAIS	Substitut.	*	582	1744	p.(Val582 Phe)						bilateral inguinal hernia	Female	Normal		Bouvattier et al. J Clin Endorinol & Metab 87: 29-32, 2002
		2 DBD		2859	c.1744G	2859G >T										
1031	CAIS	Substitut.	*	582	1744	p.(Val582 Phe)							Female	Normal		Hughes et al., The Lancet 380:1419-1428, 2012
		2 DBD		2859	c.1744G	2859G >T										
0719	PAIS	Substitut.		582	1744	p.(Val582 Leu)						niece also affected	Female	Ambiguous	pos	Ledig et al: Horm Res 63: 263-269, 2005
		2 DBD		2859	c.1744G	2859G >C										
1032	PAIS	Substitut.	*	582	1744	p.(Val582 Leu)							Female	Ambiguous		Hughes et al., The Lancet 380:1419-1428, 2012
		2 DBD		2859	c.1744G	2859G >C										
1033	PAIS	Substitut.	*	582	1745	p.(Val582 Gly)							Female	Ambiguous		Hughes et al., The Lancet 380:1419-1428, 2012
		2 DBD		2860	c.1745T	2860T >G										

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0047	PAIS	Substitut.		583	1748	p.(Phe583 Ser)			zero				Female	Ambiguous		Hiort et al. Hum Mol Genet 3: 1163-1166 1994
		2 DBD		2863	c.1748T	2863T >C										
0046	PAIS	Substitut.		583	1748	p.(Phe583 Tyr)			normal	normal		Reduced transcription & DNA binding	Female	Ambiguous	pos	Imasaki et al. Mol & Cell Endocrinol 120: 15 -24, 1996
		2 DBD		2863	c.1748T	2863T >A										
0045	CAIS	Deletion	*	584	1750	p.(Phe584 del)	22	23	low	normal		3 nt del - p.Phe584del	Female	Normal	neg	Beitel et al. Hum Mol Genet, 3: 21, 1994
		2 DBD		2865	c.1750_1752	2865_2867 delTTC										
0442	CAIS	Deletion		584	1750	p.(Phe584 del)			normal	normal		3 nt del - p.Phe584del	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		2 DBD		2865	c.1750_1752	2865_2867 delTTC										
0597	CAIS	Deletion		584	1750	p.(Phe584 del)						3 nt del - p.Phe584del	Female	Normal		Scheiber et al. J Pedatric Endocrinol & Metab. 16: 367-373,
		2 DBD		2865	c.1750_1752	2865_2867 delTTC										
0974	CAIS	Substitut.		584	1752	p.(Phe584 Leu)	29	17				Mother & 2 sisters heterozygous carriers 1 sister & 1 aunt affct	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		2 DBD		2867	c.1752C	2867C >G										
0048	CAIS	Substitut.		586	1757	p.(Arg586 Lys)							Female	Normal		Sultan et al. J Steroid Biochem & Mol Biol: 46
		2 DBD		2872	c.1757G	2872G >A										
0488	Prostate Cancer	Substitut.		587	1760	p.(Ala587 Val)						Somatic mutation	Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000
		2 DBD		2875	c.1760C	2875C >T										
0877	CAIS	Substitut.	*	587	1760	p.(Ala587 Asp)			normal			Zero transactivation activity	Female	Normal	pos	Rajender et al. Fert Steril 91: 933.e23-28, 2009
		2 DBD		2875	c.1760C	2875C >A										
0490	Prostate Cancer	Substitut.		588	1762	p.(Ala588 Ser)						Somatic mutation	Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000
		2 DBD		2877	c.1762G	2877G >T										
0049	CAIS	Deletion							zero			Similar 2-8 deletion in 2 different families	Female	Normal		Jakubiczka et al. Human Mutation 9: 57 -61, 1997
		2-8														
0050	CAIS	Deletion	*						high	normal		Produces internally deleted protein	Female	Normal	pos	Quigley et al. Mol Endocrinol 6: 1103, 1992
		3 DBD														
0051	CAIS	Deletion											Female	Normal	pos	Hiort et al. Am J Med Genet. 63: 218-22, 1996
		3 DBD														
0443	CAIS	Deletion							zero				Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		3 DBD														
0444	CAIS	Deletion											Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		3 DBD														
0052	CAIS	Substitut.	*	591	1771	p.(Lys591 *)			zero				Female	Normal		Marcelli et al: Mol Endocrinol 4: 1105, 1990
		3 DBD		2886	c.1771A	2886A >T										
0975	CAIS	Substitut.		594	1782	p.(Tyr594 *)	18	18					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		3 DBD		2897	c.1782C	2897C >A										
0887	Prostate Cancer	Substitut.		595	1783	p.(Leu595 Met)						Q58L,Q86del,Q262*, E666D, AR23. Bical treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		3 DBD		2898	c.1783C	2898C >A										
0053	PAIS	Substitut.	**	597	1789	p.(Ala597 Thr)			normal	normal		Found in 2 unrelated fam. Abolishes dimerization	Male	Ambiguous	pos	Gast et al. Mol & Cell Endocrinol 111: 93-98, 1995
		3 DBD		2904	c.1789G	2904G >A										
0434	PAIS	Substitut.	*	597	1789	p.(Ala597 Thr)			normal	normal		Somatic mosaicism	Male	Ambiguous		Holterhus et al. Pediatric Res 46: 684 -690, 1999
		3 DBD		2904	c.1789G	2904G >A										
0510	PAIS	Substitut.	**	597	1789	p.(Ala597 Thr)			normal			partial transactivation in COS cells	Male	Ambiguous		Giwercman et al. Hormone Research 53: 83-88, 2000
		3 DBD		2904	c.1789G	2904G >A										
0710	PAIS	Substitut.	*	597	1789	p.(Ala597 Thr)			high				Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
		3 DBD		2904	c.1789G	2904G >A										
0054	PAIS	Substitut.	*	598	1792	p.(Ser598 Gly)			normal	normal		High disso rate. Also has p.R618P 2968G>C mut.	Female	Ambiguous		Zoppi et al. Mol Endocrinol 6: 409-415, 1992
		3 DBD		2907	c.1792A	2907A >G										
0390	PAIS	Substitut.		598	1793	p.(Ser598 Thr)						Servere hypospadia and cryptorchidism	Male	Ambiguous		Nordenskjold et al Urological Res. 27: 49 -55, 1999
		3 DBD		2908	c.1793G	2908G >C										
0609	PAIS	Substitut.		598	1794	p.(Ser598 Arg)						Newborn treated with 1% DHT locally - normal penile size	Male	Ambiguous		Giwercmann et al. Horm Res 61: 58-62, 2004
		3 DBD		2909	c.1794C	2909C >A										

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0610	Normal	Substitut.		598	1794	p.(Ser598 Arg)						Adult had bilateral cryptorchadism in childhood	Male	Normal		Giwercmann et al. Horm Res 61: 58-62, 2004
		3 DBD		2909	c.1794C	2909C >A										
0959	E006AA Prost Can cell line	Substitut.		598	1792	p.(Ser598 Gly)						Dominant-negative loss of function mut AR gene amplificat	Male	Normal		D'Antonio et al. PLOS One 5: e11475, 2010
		3 DBD		2907	c.1792A	2907A >G										
0720	CAIS	Deletion		600	1799	p.(Asn600 fs)						1 nt deletion causing frameshift & stop at codon 625	Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
		3 DBD		2914	c.1799A	2914A delA										
0055	CAIS	Substitut.		602	1805	p.(Cys602 Phe)							Female	Normal	pos	Baldazzi et al. Hum Mol Genet 3: 1169-70 1994
		3 DBD		2920	c.1805G	2920G >T										
0946	PAIS	Substitut.	*	602	1805	p.(Cys602 Ser)						40% loss of ligand-binding & 70% loss of transacrivation	Female	Ambiguous		Singh et al. J Steroid Biochem Mol Biol 2010
		3 DBD		2920	c.1805G	2920G >C										
1075	CAIS	Substitut.	*	602	1805	p.(Cys602 Tyr)							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		3 DBD		2920	c.1805G	2920G >A										
0362	PAIS	Substitut.		603	1807	p.(Thr603 Pro)	24					testis located in ingunal canal. Same family as 0831	Male	Ambiguous	pos	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
		3 DBD		2922	c.1807A	2922A >C										
0831	PAIS	Substitut.		603	1807	p.(Thr603 Pro)						Same family as 0362	Male	Ambiguous	pos	Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
		3 DBD		2922	c.1807A	2922A >C										
0942	PAIS	Substitut.		604	1811	p.(Ile604 Asn)			zero			Somatic mosaicism - no transcriptional activity				Elfferich et al. Sexual Development 3: 237 -244, 2009
		3 DBD		2926	c.1811T	2926T >A										
1034	CAIS	Substitut.		604	1811	p.(Ile604 Ser)						Twins	Male	Normal		Hughes et al., The Lancet 380:1419-1428, 2012
		3 DBD		2926	c.1811T	2926T >G										
0056	PAIS	Substitut.		605	1813	p.(Asp605 Tyr)							Male	Ambiguous		Hiort et al: Hum Mol Genet 3: 1163-1166 1994
		3 DBD		2928	c.1813G	2928G >T										
0598	PAIS	Substitut.		605	1813	p.(Asp605 Tyr)							Female	Ambiguous	pos	Scheiber et al. J Ped Endocrinol & Metab. 16: 367-373, 2003
		3 DBD		2928	c.1813G	2928G >T										
1074	PAIS	Substitut.		605	1813	p.(Asp605 Asn)										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		3 DBD		2928	c.1813G	2928G >A										
0848	CAIS	Substitut.	*	608	1822	p.(Arg608 *)						Diag at 1 Mo-Wolffian derivatives on rt side only	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
		3 DBD		2937	c.1822C	2937C >T										
0057	CAIS	Substitut.	*	608	1822	p.(Arg608 *)			zero				Female	Normal		Brown et al. Eur J Pediatr (Suppl 2) 152. S62, 1993
		3 DBD		2937	c.1822C	2937C >T										
0511	CAIS	Substitut.	*	608	1822	p.(Arg608 *)			zero				Female	Normal		Giwercman et al. Hormone Research 53: 83-88, 2000
		3 DBD		2937	c.1822C	2937C >T										
0702	CAIS	Substitut.	*	608	1822	p.(Arg608 *)						1 affected sibling	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
		3 DBD		2937	c.1822C	2937C >T										
0770	MAIS	Substitut.	*	608	1823	p.(Arg608 Gln)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		3 DBD		2938	c.1823G	2938G >A										
0677	PAIS	Substitut.	*	608	1823	p.(Arg608 Gln)								Ambiguous		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
		3 DBD		2938	c.1823G	2938G >A										
0393	PAIS	Substitut.	*	608	1823	p.(Arg608 Gln)						Germ cell tumour - in undescended testis	Female	Normal		Chen et al. Human Reproduction 14: 664 -670, 1999
		3 DBD		2938	c.1823G	2938G >A										
0347	PAIS	Substitut.	*	608	1823	p.(Arg608 Gln)						Patient successfully treated with testostertone	Male	Ambiguous		Weidemann et al. J Clin Endocrinol & Metab 83: 1173-1176, 1998
		3 DBD		2938	c.1823G	2938G >A										
0060	PAIS	Substitut.	*	608	1823	p.(Arg608 Gln)							Female	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
		3 DBD		2938	c.1823G	2938G >A										
0059	PAIS	Substitut.	**	608	1823	p.(Arg608 Gln)			normal	normal			Male	Ambiguous	pos	Weidemann et al. Clin Endocrinology 45: 733 -739, 1996
		3 DBD		2938	c.1823G	2938G >A										
0058	PAIS and breast cancer	Substitut.	*	608	1823	p.(Arg608 Gln)							Male	Ambiguous	pos	Wooster et al. Nat Genet 2: 132, 1992
		3 DBD		2938	c.1823G	2938G >A										
0847	CAIS	Substitut.		608	1822	p.(Arg608 *)						Diag at 1Mo Bilateral gonadectomy at 16y	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
		3 DBD		2937	c.1822C	2937C >T										

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0061	PAIS	Substitut.		609	1826	p.(Arg609 Lys)			normal	normal			Male	Ambiguous		Saunders et al. Clin Endocrinol 37: 214, 1992
		3 DBD		2941	c.1823G	2941G >A										
0062	PAIS and breast cancer	Substitut.		609	1826	p.(Arg609 Lys)			normal	normal			Male	Ambiguous		Lobaccaro et al. Hum Mol Genet, 2: 1799, 1993
		3 DBD		2941	c.1823G	2941G >A										
0322	PAIS	Substitut.		609	1826	p.(Arg609 Lys)			normal	normal		Defective nuclear localization	Male	Ambiguous		Tincello et al. Clinical Endocrinology 46: 497 -506, 1997
		3 DBD		2941	c.1823G	2941G >A										
0352	PAIS	Substitut.		609	1826	p.(Arg609 Lys)							Male	Ambiguous	pos	Hiort et al. J Pediatrics 132: 939- 943, 1998
		3 DBD		2941	c.1823G	2941G >A										
0481	PAIS	Substitut.		609	1826	p.(Arg609 Lys)			normal	high						Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		3 DBD		2941	c.1823G	2941G >A										
0412	CAIS	Deletion		609	1825	p.(Arg609 fs)						Mullerian ducts pres. 5nt del frameshift & stop in codon 621	Female	Normal		Chen et al. Fertility & Sterility 72: 170-173, 1999
		3 DBD		2940	c.1825_1829	2940_2944 delAGGAA										
0956	PAIS	Substitut.		609	1826	p.(Arg609 Lys)							Male	Ambiguous	neg	Wu et al. Fertility & Sterility 93: 2076, e1-4, 2010
		3 DBD		2941	c.1826G	2941G >A										
0063	PAIS	Substitut.	*	611	1832	p.(Asn611 Thr)			normal	low	*		Male	Ambiguous		Weidemann et al. Clin Endocrinology 45: 733 -739, 1996
		3 DBD		2947	c.1832A	2947A >C										
0721	CAIS	Insertion		611	1831	p.(Asn611 fs)							Female	Normal	neg	Ledig et al. Horm Res 63: 263-269, 2005
		3 DBD		2946	c.1831_1832	2946_2947 insA										
0496	CAIS	Substitut.		612	1835	p.(Cys612 Tyr)							Female	Normal		Mockel et al. Geburtshi. und Frauen. 60: 232 -234, 2000
		3 DBD		2950	c.1835G	2950G >A										
0722	CAIS	Substitut.		615	1835	p.(Cys615 Tyr)							Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
		3 DBD		2959	c.1835G	2959G >A										
1076	CAIS	Substitut.		616	1835	p.(Arg616 Cys)							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		3 DBD		2961	c.1846C	2961C >T										
0512	CAIS	Substitut.	*	616	1846	p.(Arg616 Gly)						no transactivation in COS cells	Female	Normal		Giwercman et al. Hormone Research 53: 83-88, 2000
		3 DBD		2961	c.1846C	2961C >G										
0065	CAIS	Substitut.	**	616	1847	p.(Arg616 His)	25	23	low	high			Female	Normal	pos	Beitel et al. Hum Mol Genet, 3: 21-27, 1994
		3 DBD		2962	c.1847G	2962G >A										
0066	CAIS	Substitut.	**	616	1847	p.(Arg616 His)			normal	normal			Female	Normal	pos	Mowszowicz et al. Mol Endocrinol 7: 861-869, 1993
		3 DBD		2962	c.1847G	2962G >A										
0067	CAIS	Substitut.	**	616	1847	p.(Arg616 His)							Female	Normal		Brown et al. Eur J Pediatr 152 (Suppl 2): S62, 1993
		3 DBD		2962	c.1847G	2962G >A										
0068	CAIS	Substitut.	**	616	1847	p.(Arg616 His)						2 affected siblings variable phenotype	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
		3 DBD		2962	c.1847G	2962G >A										
0348	CAIS	Substitut.	*	616	1847	p.(Arg616 His)							Female	Normal		Cabral et al. Brazilian J Mol & Biol Res. 31: 775-778, 1998
		3 DBD		2962	c.1847G	2962G >A										
0353	CAIS	Substitut.	*	616	1847	p.(Arg616 His)							Female	Normal		Hiort et al. J Pediatrics 132: 939- 943, 1998
		3 DBD		2962	c.1847G	2962G >A										
0354	CAIS	Substitut.	*	616	1847	p.(Arg616 His)							Female	Normal		Hiort et al. J Pediatrics 132: 939- 943, 1998
		3 DBD		2962	c.1847G	2962G >A										
0069	PAIS	Substitut.	*	616	1847	p.(Arg616 His)							Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
		3 DBD		2962	c.1847G	2962G >A										
0654	CAIS	Substitut.	*	616	1847	p.(Arg616 His)						Wolffian remnants present - sister of 655	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		3 DBD		2962	c.1847G	2962G >A										
0655	CAIS	Substitut.	*	616	1847	p.(Arg616 His)						Sister of 654	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		3 DBD		2962	c.1847G	2962G >A										
0069	MAIS	Substitut.	*	616	1847	p.(Arg616 His)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		3 DBD		2962	c.1847G	2962G >A										
0070	PAIS	Substitut.		616	1847	p.(Arg616 Pro)							Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
		3 DBD		2962	c.1847G	2962G >C										

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0445 CAIS	Substitut.	3 DBD		616 2962	1847 c.1847G	p.(Arg616 Pro) 2962G >C			normal	high			Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0064 CAIS	Deletion	3 DBD		616 2961	1846 c.1846_1848	p.(Arg616 del) 2961-2963 delCGT	27	23	normal	normal		3 nt del - p.R616del	Female	Normal		Beitel et al. Hum Mol Genet, 3: 21-27, 1994
1025 CAIS	Substitut.	3 DBD		616 2961	1846 c.1846C	p.(Arg616 Ser) 2961C >A						mother is a heterozygous carrier	Female	Normal	pos	Sharma et al. Fertil Steril 95: 804, e19-21, 2011.
0071 PAIS	Substitut.	3 DBD	*	617 2965	1850 c.1850T	p.(Leu617 Arg) 2965T >G			normal	normal			Female	Ambiguous	pos	De Bellis et al. J Clin Endocrinol Metab, 78: 513, 1994
0072 CAIS	Substitut.	3 DBD		617 2965	1850 c.1850T	p.(Leu617 Pro) 2965T >C							Female	Normal		Mebarki et al. 75th US Endo Soc Meeting, Abstr 602,1993
0073 CAIS	Substitut.	3 DBD	*	617 2965	1850 c.1850T	p.(Leu617 Pro) 2965T >C			normal	normal			Female	Normal		Lobaccaro et al. Mol Cell Endorinol, 5: 137 -147, 1996
1110 CAIS	Substitut.	3 DBD		618 2967	1852 c.1852C	p. (Arg 618 * 2967C >T 2968 delG						Persistence of Wolffian derivatives	Female	Normal		Liu et al. J Androl 33:357-360, 20123
0074 PAIS	Substitut.	3 DBD	*	618 2968	1853 c.1853G	p.(Arg618 Pro) 2968G >C			normal	normal			Female	Ambiguous	pos	Marcelli et al. J Clin Invest. 87: 1123, 1991
0075 PAIS	Substitut.	3 DBD	*	618 2968	1853 c.1853G	p.(Arg618 Pro) 2968G >C			normal	normal	high	Mutation also at 598	Female	Normal		Zoppi et al. Mol Endocrinol 6: 409, 1992
0431 Prostate cancer	Substitut.	3 DBD	*	620 2974	1859 c.1859G	p.(Cys620 Tyr) 2974G >A			low	high		Inactive transcription Does not bind DNA somatic mutation	Male	Normal		Nazereth et al. Mol Endocrinol 13: 2065 -2075, 1999
0976 CAIS	Substitut.	3 DBD		620 2973	1858 c.1858T	p.(Cys620 Arg) 2973T >C	20	18				Mother heterozygous carrier, 1 affected sister	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0491 Prostate cancer	Substitut.	3 DBD		620 2974	1859 c.1859G	p.(Cys620 Tyr) 2974G >A						Somatic mutation	Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000
0076 CAIS	Deletion	3-8											Female	Normal		Brown et al, Eur J Pediatr (Suppl 2) 152: S62, 1993
0711 PAIS	Substitut.	4		630 3003	1888 c.1888C	p.(Arg630 Trp) 3003C >T			normal			40% higher transactivation than WT impaired N/C	Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
0306 Prostate cancer	Substitut.	4		630 3004	1889 c.1889G	p.(Arg630 Gln) 3004G >A						1 of 6 of hormone-independent D2 patients- somatic mut	Male	Normal		Wang et al. Japanese J of Urology 88: 550-556 1997
0079 Prostate cancer	Substitut.	4		631 3007	1892 c.1892A	p.(Lys631 Thr) 3007A >C						Also p.K718E mut.3267A>G,+ silent	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0077 MAIS	Deletion	4 LBD										Azoospermia	Male	Normal	neg	Aiken et al. Am J Obs & Gyn . 165: 1891 -1894, 1991
1028 CAIS	Deletion	4 LBD		637 3025	1910 c.1910	p.(Asn637 fs) 3025I delA						leads to p.Asn637Ile & Leu638*. Mother heterozygous	Female	Normal	pos	Yue et al. Zhonghua 27: 631-633, 2010
0400 CAIS	Substitut.	4 LBD		641 3036	1921 c.1921C	p.(Gln641 *) 3036C >T			zero				Female	Normal		Yaegashi et al. Tohoku J of Exp Med 187: 263 -272, 1999
0429 CAIS	Substitut.	4 LBD		641 3036	1921 c.1921C	p.(Gln641 *) 3036C >T			zero			also p.W752* mut. 3371G>A, 47XXY, Muts on both X's	Female	Normal		Uehara et al. Am J Med Genet 86: 107-111, 1999
0613 Prostate cancer	Substitut.	4 LBD	*	641 3036	1921 c.1921C	p.(Gln641 *) 3036C >T						also p.T 878A. transactiv androgen-independent ~275x induction	Male	Normal		Ceraline et al. Int. J. Cancer 108: 152-157, 2004
0080 PAIS	Substitut.	4 LBD		646 3052	1937 c.1937C	p.(Ala646 Asp) 3052C >A							Male	Ambiguous		Hiort et al. Am J Med Genet 63: 218-222, 1996
0334 Normal	Substitut.	4 LBD		646 3052	1937 c.1937C	p.(Ala646 Asp) 3052C >A							Male	Normal		Nordenskjold et al. Human Mutation. 11: 339, 1998
0617 CAIS	Substitut.	4 LBD		646 3052	1937 c.1937C	p.(Ala646 Asp) 3052C >A	27		low			Also p.R753Q	Female	Normal		MacLean et al. Hum Mutat 23: 287, 2004
0772 MAIS	Substitut.	4 LBD		646 3052	1937 c.1937C	p.(Ala646 Asp) 3052C >A						male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006

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0812	PAIS	Substitut.	*	646	1937	p.(Ala646 Asp)	28					Long poly Q and short poly G contribute to PAIS	Male	Ambiguous		Werner et al. J Clin Endocrinol Metab 91: 3515-3520, 2006
		4 LBD		3052	c.1937C	3052C >A										
0813	PAIS	Substitut.	*	646	1937	p.(Ala646 Asp)	30					Long poly Q and short poly G contribute to PAIS	Male	Ambiguous		Werner et al. J Clin Endocrinol Metab 91: 3515-3520, 2006
		4 LBD		3052	c.1937C	3052C >A										
1078	PAIS	Substitut.		646	1937	p.(Ala646 Asp)	30									Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		4 LBD		3052	c.1937C	3052C >A										
0081	Prostate cancer	Substitut.		648	1943	p.(Ser648 Asn)						Also p.G725D, L881Q & A897T mut Somatic mutations	Male	Normal		Taplin et al. New England J Med 332: 1393-1398, 1995
		4 LBD		3058	c.1943G	3058G >A										
1020	Premature ovarian failure	Substitut.		650	1948	p.(Thr650 Ala)						Patient had menopause at 28	Female	Normal		Panda et al. Gynecol Endocrinol 27:1-7, 2011
		4 LBD		3063	c.1948A	3063A >G										
0576	Prostate cancer	Substitut.		650	1950	p.(Thr650 Thr)						Somatic mut. -CaP Poorly diff, Stage B2 +Estramustine treat	Male	Normal		Segawa et al. Int J of Urology 9: 545-553, 2002
		4 LBD		3065	c.1950C	3065C >T										
0784	MAIS	Substitut.		651	1951	p.(Ser651 Gly)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		4 LBD		3066	c.1951A	3066A >G										
0827	MAIS	Substitut.	*	651	1951	p.(Ser651 Gly)	23	18				Hypoandrogenism, scrotal hypoplsia hypospematogenesis	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
		4 LBD		3066	c.1951A	3066A >G										
0555	PAIS	Substitut.		654	1960	p.(Glu654 Lys)	20					Also in family with CAH with no androgen insensitivity	Male	Ambiguous		Lundberg et al. J Clin Endocrinol Metab 87: 2023-2028, 2002
		4 LBD		3075	c.1960G	3075G >A										
0517	CAIS	Substitut.		658	1972	p.(Gln658 *)							Female	Normal		Chavez et al. Clin Genet 59: : 185-188, 2001
		4 LBD		3087	c.1972C	3087C >T										
1021	Premature ovarian failure	Substitut.		658	1972	p.(Gln658 Lys)						Patient had menopause at 17	Female	Normal		Panda et al. Gynecol Endocrinol 27: 1-7, 2011
		4 LBD		3087	c.1972C	3087C >A										
0082	PAIS	Substitut.		665	1994	p.(Ile665 Asn)	22	22	low		norm					Pinsky et al. Clin Inv Med 15: 456, 1992
		4 LBD		3109	c.1994T	3109T >A										
1079	PAIS	Substitut.		665	1994	p.(Ile665 Asn)										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		4 LBD		3109	c.1994T	3109T >C										
0937	Prostate cancer	Substitut.		666		p.(Glu666 Asp)						Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		4 LBD														
1083	CAIS	Substitut.		671	2011	p.(Gln *)							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		4 LBD		3126	c.2011	3126G >T										
0083	Prostate cancer	Substitut.		671	2012	p.(Gln671 Arg)						Also p.S792P 3489T>C mut. Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
		4 LBD		3127	c.2012A	3127A >G										
0084	PAIS	Substitut.		672	2015	p.(Pro672 His)							Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
		4 LBD		3130	c.2015C	3130C >A										
0943	CAIS	Substitut.	*	672	2014	p.(Pro672 Ser)						Reduced transactivat no change in DNA or FxxLF binding	Female	Normal		Elfferich et al. Sexual Development 3: 237 -244, 2009
		4 LBD		3129	c.2014C	3129C >T										
0085	Prostate cancer	Substitut.		673	2018	p.(Ile673 Thr)						Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
		4 LBD		3133	c.2018T	3133T >C										
1079	PAIS	Substitut.		674	2021	p.(Phe674 Cys)										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		4 LBD		3136	c.2021T	3136T >G										
0086	CAIS	Substitut.		678	2033	p.(Leu678 Pro)			zero				Female	Normal	pos	Belsham et al. Human Mutation 5: 28-33, 1995
		4 LBD		3148	c.2033T	3148T >C										
1080	PAIS	Substitut.		679	2036	p.(Glu679 Gly)										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		4 LBD		3151	c.2036A	3151A >G										
0723	CAIS	Substitut.		681	2042	p.(Ile681 Thr)						Affected twin sister	Female	Normal	pos	Ledig et al. Horm Res 63: 263-269, 2005
		4 LBD		3157	c.2042T	3157T >C										
1005	PAIS	Substitut.		681	2042	p.(Ile681 Asn)	21	4	normal	high		sister , cousin and 2 great aunts affected	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		4 LBD		3157	c.2042T	3157T >A										
0087	CAIS	Substitut.		682	2044	p.(Glu682 Lys)							Female	Normal		Hiort et al. J Clin Endocrinol Metab 77: 262-266, 1993
		4 LBD		3159	c.2044G	3159G >A										

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0394	CAIS	Substitut.		682	2044	p.(Glu682 Lys)						Germ cell tumour in undescended testis	Female	Normal		Chen et al. Human Reproduction 14: 664 -670, 1999
		4 LBD		3159	c.2044G	3159G >A										
0618	CAIS	Substitut.		682	2044	p.(Glu682 *)	21		v low				Female	Normal		MacLean et al. Hum Mutat 23: 287, 2004
		4 LBD		3159	c.2044G	3159G >T										
0619	CAIS	Substitut.		682	2044	p.(Glu682 *)	21		low				Female	Normal		MacLean et al. Hum Mutat 23: 287, 2004
		4 LBD		3159	c.2044G	3159G >T										
947	CAIS	Substitut.		682	2046	p.(Glu682 Asp)						Mother a carrier- 3 affected siblings	Female	Normal	pos	Wu et al. Zhonghua Yi Xue Yi Chuan Xue Za Zhi 26: 606-609 2009
		4 LBD		3161	c.2046G	3161G >T										
0534	PAIS	Substitut.		683	2047	p.(Pro683 Thr)			low				Female	Ambiguous		Chavez et al. J Hum Genet. 46: 560-565, 2001
		4 LBD		3162	c.2047C	3162C >A										
0724	CAIS	Substitut.		683	2047	p.(Pro683 Ala)			low			aunt, 2 sisters and neice affected	Female	Normal	pos	Ledig et al. Horm Res 63: 263-269, 2005
		4 LBD		3162	c.2047C	3162C >G										
0089	Prostate cancer	Substitut.		684	2051	p.(Gly684 Ala)						Somatic mutation - transactivation normal	Male	Normal		Koivisto et al. Cancer Research 57: 314-319, 1997
		4 LBD		3166	c.2051G	3166G >C										
0090	CAIS	Substitut.		685	2053	p.(Val685 Ile)			zero				Female	Normal		Mebarki et al. 75th US Endo Soc Meeting, Abstr 602, 1993
		4 LBD		3168	c.2053G	3168G >A										
0945	CAIS	Duplicat.		686	2055	p.(Val686 fs)						2 nt duplicat. causing frameshift & stop in Codon 788	Female	Normal	pos	Rong et al. Eur J Obst & Gynecol & Reprod Biol 148: 53-55, 2010
		4 LBD		3170	p.2055_2056	3170 _3171 dupAG										
0091	PAIS	Substitut.		687	2059	p.(Cys687 Arg)							Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
		4 LBD		3174	c.2059T	3174T >C										
1081	PAIS	Substitut.		688	2063	p.(Ala688 Asp)										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		4 LBD		3178	c.2063C	3178C >A										
0092	PAIS	Substitut.		688	2063	p.(Ala688 Val)							Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
		4 LBD		3178	c.2063C	3178C >T										
0093	CAIS	Substitut.		689	2066	p.(Gly689 Glu)						de novo mutation	Female	Normal	neg	Hiort et al. J Pediatrics 132: 939-943, 1998
		4 LBD		3181	c.2066G	3181G >A										
0446	CAIS	Substitut.		689	2065	p.(Gly689 *)			zero				Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		4 LBD		3180	c.2065G	3180G >T										
0575	CAIS	Substitut.	*	690	2069	p.(His690 Pro)			v low	low	high *	v. low transactivation activity	Female	Normal		Rosa et al. J Clin Endocrinol Metab 87. 4378-4382, 2002.
		4 LBD		3184	c.2069A	3184A >C										
0094	PAIS	Deletion		691	2071	p.(Asp691 del)										Schwartz et al. Horm Res 41: 117 Abstr 244, 1994
		4 LBD		3186	c.2071_2073	3186_3188 delGAC										
0656	CAIS	Substitut.	*	691	2072	p.(Asp691 Val)				high		Epididymis & Vas deferens present - v low transactivation	Female	Normal		Tadokoro et al. Clinical Endocrinology 71: 253 -260, 2009
		4 LBD		3187	c.3187A	3187A >T										
1082	PAIS	Substitut.		691	2073	p.(Asp691 Glu										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
		4 LBD		3188	c.2073C	3188C										
0095	CAIS	Deletion		693	2077	p.(Asn693 del)			normal	high	*	Three nucleotide deletion	Female	Normal		Batch et al. Hum Mol Genet 1: 497-503, 1992
		4 LBD		3192	c.2077_2079	3192 _3194 delAAC										
0704	PAIS	Deletion		693	2077	p.(Asn693 del)			normal	high		3 nt deletion. 3 affected siblings . variable phenotype	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
		4 LBD		3192	c.2077_2079	3192_3194 delAAC										
0849	CAIS	Substitut.		694	2080	p.(Gln694 *)						Diag at 17y- Bilateral gonadectomy 17y	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
		4 LBD		3195	c.2080C	3195C >T										
0096	CAIS	Substitut.	*	696	2086	p.(Asp696 His)			low				Female	Normal	neg	Ris-Stalpers et al. Mol Endocrinol 5: 1562, 1991
		4 LBD		3201	c.2086G	3201G >C										
0097	CAIS	Substitut.	**	696	2086	p.(Asp696 Asn)			normal	normal	high	mutation found in two unrelated families	Female	Normal	pos	Ris-Stalpers et al. Mol Endocrinol 5: 1562, 1991
		4 LBD		3201	c.2086G	3201G >A										
0098	PAIS	Substitut.	*	696	2086	p.(Asp696 Asn)						de novo mutation	Female	Ambiguous		Hiort et al. J Pediatrics 132: 939- 943, 1998
		4 LBD		3201	c.2086G	3201G >A										
0657	CAIS	Substitut.	*	696	2086	p.(Asp696 Asn)						Wolffian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		4 LBD		3201	c.2086G	3201G >A										

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0773 MAIS	Substitut.	4 LBD	*	696 3201	2086 c.2086G	p.(Asp696 Asn) 3201G >A						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
0977 CAIS	Substitut.	4 LBD	*	696 3201	2086 c.2086G	p.(Asp696 Asn) 3201G >A						Diag at 2 wk bilateral gonadectomy at 14yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0978 CAIS	Substitut.	4 LBD	*	696 3201	2086 c.2086G	p.(Asp696 Asn) 3201G >A	22	17				Mother & 1 cousin hetrozyg, 1 cousin & 1 aunt affect, sister wt	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0977 CAIS	Substitut.	4 LBD		696 3201	2086 c.2086G	p.(Asp696 Tyr) 3201G >T	19	18				Mother heterozygous carrier	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0335 CAIS	Substitut.	4 LBD		696 3202	2087 c.2087A	p.(Asp696 Val) 3202A >T	21					mutation found in two siblings	Female	Normal	pos	Dork et al. Human Mutation 11: 337-339, 1998
1053 CAIS	Substitut.	4 LBD		698 3209	2094 c.2094T	p.(Phe698 Leu) 3209T >G						24 yr. Inguinal cryptorchism	Female	Normal		Ning et al. Urology 80: 216-218, 2012
0447 CAIS	Substitut.	4 LBD		701 3216	2101 c.2101T	p.(Leu701 Met) 3216T >A			zero			Epididymous and Vas deferns present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0725 CAIS	Deletion	4 LBD		701 3216	2101 c.2101_2114	p.(Leu701 fs) 3216_3228 del						13 nt del causing frameshift. sister & mother carr, neice aff	Female	Normal	pos	Ledig et al. Horm Res 63: 263-269, 2005
1084 CAIS	Substitut.	4 LBD		701 3218	2103 c.2103G	p.(Leu701 Phe) 3218G							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0448 CAIS	Substitut.	4 LBD		702 3209	2094 c.2094C	p.(Leu702 Phe) 3209C >T							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0518 PAIS	Substitut.	4 LBD		702 3219	2104 c.2104C	p.(Leu702 Ile) 3219C >A										Chavez et al. Clin Genet 59: : 185-188, 2001
0099 Prostate cancer	Substitut.	4 LBD		702 3220	2105 c.2105T	p.(Leu702 His) 3220T >A						Somatic mutation	Male	Normal		Suzuki et al. J Steroid Biochem Molec Biol 46: 759, 1993
0326 Prostate cancer	Substitut.	4 LBD		702 3220	2105 c.2105T	p.(Leu702 His) 3220T >A						Somatic mutation	Male	Normal		Watanabe et al. Jpn J Clin Oncol 27: 389 -393, 1997
0408 MDA PCa-Za	Substitut.	4 LBD		702 3220	2105 c.2105T	p.(Leu702 His) 3220T >A			normal	low		Som. mut. Prostate cancer cell line. Also has T878A	Male	Normal		Zao et al. J of Urology 162: 2192-2199, 1999
0100 CAIS	Substitut.	4 LBD		703 3222	2107 c.2107T	p.(Ser703 Ala) 3222T >G			zero				Female	Normal		Pinsky et al. Clin Inv Med 15: 456-472, 1992
0101 PAIS	Substitut.	4 LBD	*	704 3225	2110 c.2110A	p.(Ser704 Gly) 3225A >G			low	high			Male	Ambiguous		Radnayr et al. J of Urology 158: 1553 -1556, 1997
0449 CAIS	Substitut.	4 LBD	*	704 3225	2110 c.2110A	p.(Ser704 Gly) 3225A >G							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0712 PAIS	Substitut.	4 LBD		704 3225	2110 c.2110A	p.(Ser704 Gly) 3225A >G				high			Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
0713 PAIS	Substitut.	4 LBD		704 3225	2110 c.2110A	p.(Ser704 Gly) 3225A >G							Female	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
0979 CAIS	Substitut.	4 LBD		704 3225	2110 c.2110A	p.(Ser704 Cys) 3225A >T	19	18	low	normal		Mother, sister, heterozygote carriers, I aunt wt	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1085 CAIS	Substitut.	4 LBD		704 3225	2110 c.2110A	p.(Ser704 Cys) 3225A >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
1108 CAIS	Substitut.	4 LBD		704 3226	2111 c.2111G	p.(Ser704 Ile) 3226G >T						Sertoli cell adenoma-persistence of Mullerian derivatives	Female	Normal		Chin et al. J Pediatr Endocrinol Metab 25:1145-1051, 2012
0102 CAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ser) 3232A >G			zero				Female	Normal		Pinsky et al. Clin Inv Med 15: 456-472, 1992
0103 CAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ser) 3232A >G			zero				Female	Normal		DeBellis et al. Mol Endocrinol 6: 1909-20, 1992
0104 CAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ser) 3232A >G						Mutation found in two unrelated families	Female	Normal		Quigley et al. Endocrine Reviews 16: 271, 1995

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0482 PAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ser) 3232A >G			normal	high						Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0633 CAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ser) 3232A >G			zero			Testes located in abdomen	Female	Normal	neg	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
0634 CAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ser) 3232A >G	22		zero			Testes located in inguinal region	Female	Normal	pos	Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
0980 CAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ser) 3232A >G	18	18	zero	zero			Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0981 CAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ser) 3232A >G	22	18				1 affected sister, 1 heterozygote sister, 2 wt sisters	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1086 CAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ser) 3232A >G							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0559 CAIS	Substitut.	4 LBD		706 3231	2116 c.2116A	p.(Asn706 Tyr) 3231A >T						Sister a carrier	Female	Normal		Sills et al. Int J Mol Med 9: 45-48, 2002
0581 CAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ile) 3232A >T						Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
0582 CAIS	Substitut.	4 LBD		706 3232	2117 c.2117A	p.(Asn706 Ile) 3232A >T						Low immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
0105 CAIS	Substitut.	4 LBD	*	708 3238	2123 c.2123T	p.(Leu708 Arg) 3238T >G							Female	Normal		Lumbroso et al. J Clin Endo & Metab 81: 1984 -1988, 1996
0078 CAIS	Deletion	4 LBD	*	708 3238	2123 c.2123_2135	p.(Leu708 fs) 3238_3250 del						13 nt deletionn (TGGGAGAGACA) causing framshift and stop at codon 784	Female	Normal	pos	Lobaccaro et al. Mol & Cellular Endocrinology 111: 21-8,1995
1087 CAIS	Substitut.	4 LBD		709 3240	2125 c.2125G	p.(Gly709 Arg 3240G >A							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0106 PAIS	Substitut.	4 LBD		709 3241	2126 c.2126G	p.(Gly709 Ala) 3241G >C							Male	Ambiguous		Hiort et al: Hum Mol Genet 3: 1163-1166 1994
0314 PAIS	Substitut.	4 LBD		709 3241	2126 c.2126G	p.(Gly709 Ala) 3241G >C						Servere hypospadias	Male	Ambiguous		Albers et al. J of Pediatrics 131: 388 -392, 1997
0107 PAIS	Substitut.	4 LBD		709 3241	2126 c.2126G	p.(Gly709 Val) 3241G >T			zero			Mother had both mut. & wt. AR genes.	Male	Ambiguous	pos	Auchus et al: 77th US Endo Soc Meeting, Abstr P1-508 1995
1023 CAIS	Substitut.	4 LBD		709 3241	2126 c.2126G	p.(Gly709 Glu) 3241G >A			low			Almost complete loss of transaction	Female	Normal		Rajender et al. J Androl 32: 193-198, 2011
0819 PAIS	Substitut.	4 LBD	*	710 3243	2128 c.2128G	p.(Glu710 Lys) 3243G >A							Female	Ambiguous		Geoget et al. Mol Endocrinol 20: 724 -734, 2006
0577 Prostate cancer	Substitut.	4 LBD		710 3245	2130 c.2130G	p.(Glu710 Glu) 3245G >A						Somatic mut. CaP Moder diff, Stage D1 + biculitamide treat	Male	Normal		Segawa et al. Int J of Urology 9: 545-553, 2002
0658 CAIS	Substitut.	4 LBD		711 3247	2132 c.2132G	p.(Arg711 Thr) 3247G >C			zero			Sister to 0450 - Epididymis & Vas deferens present	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0450 CAIS	Substitut.	4 LBD		711 3247	2132 c.2132G	p.(Arg711 Thr) 3247G >C			zero			Sister to 0658- Epidiymis & Vas deferens present	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0525 PAIS	Substitut.	4 LBD	*	712 3249	2134 c.2134C	p.(Gln712 Glu) 3249C >G			v low			altered AR specificity 2x increased affinity for E2.	Female	Ambiguous	pos	Lumbroso et al. 83rd US Endo Soc Meeting, Abstr P2-29, 2001
0535 PAIS	Substitut.	4 LBD	*	712 3249	2134 c.2134C	p.(Gln712 Glu) 3249C >G			normal				Female	Ambiguous	pos	Chavez et al. J Hum Genet. 46: 560-565, 2001
0828 CAIS	Deletion	4 LBD		712 3250	2135 c.2135	p.(Gln712 fs) 3250 delA						1 nt del-frameshift & stop in codon 788- 3 affected aunts	Female	Normal	pos	Galani et al. Fertility & Sterility 90: 2008.e1 -2008.e4, 2008
0578 Prostate cancer	Substitut.	4 LBD		712 3251	2136 c.2136G	p.(Gln712 Gln) 3251G >A						Somatic mut. CaP Moder diff, Stage D2 + leuprolide treat	Male	Normal		Segawa et al. Int J of Urology 9: 545-553, 2002
0108 PAIS	Substitut.	4 LBD	*	713 3252	2137 c.2137C	p.(Leu713 Phe) 3252C >G			normal	high		Phenotypic diversity brother of 505& 506 Testost-induced	Male	Ambiguous	pos	Hiort et al. Am J Med Genet. 63: 218-222, 1996

Accession # / Phenotype	Mutation type	Pathogenicity proven: Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0505	PAIS	Substitut.	*	4 LBD	713	2137	p.(Leu713 Phe)			normal	high	Phenotypic diversity brother of 108& 506 Testost-induced	Male	Ambiguous	pos	Hiort et al. J Clin Endocrinol Metab 85: 3245-3250, 2000
					3252	c.2137C	3252C >G									
0506	PAIS	Substitut.	*	4 LBD	713	2137	p.(Leu713 Phe)			normal	high	Phenotypic diversity brother of 505& 108 Testost-induced	Male	Ambiguous	pos	Hiort et al. J Clin Endocrinol Metab 85: 3245-3250, 2000
					3252	c.2137C	3252C >G									
0507	PAIS	Substitut.	*	4 LBD	713	2137	p.(Leu713 Phe)			normal	high	Phenotypic diversity uncle of 108,505,506 Testost-induced	Male	Ambiguous	pos	Hiort et al. J Clin Endocrinol Metab 85: 3245-3250, 2000
					3252	c.2137C	3252C >G									
0109	Prostate cancer	Substitut.	**	4 LBD	716	2146	p.(Val716 Met)			normal		Somatic mutation. Receptor showed a gain in function	Male	Normal		Culig et al. Mol Endocrinol 7: 1541-1550 1993
					3261	c.2146G	3261G >A									
0110	Prostate cancer	Substitut.	*	4 LBD	716	2146	p.(Val716 Met)			normal		Somatic mutation. Receptor showed a gain in function	Male	Normal		Bubley et al 87th Am Assoc Cancer Res Meet Abstr. 1680, 1996
					3261	c.2146G	3261G >A									
0678	CAIS	Substitut.	*	4 LBD	716		p.(Val716 *)					bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0792	PAIS	Substitut.		4 LBD	719	2155	p.(Trp719 Arg)					Somatic mosaic 2/3 mutant to 1/3 wt				Mueller et al. Hum Genet 119: 680, 2006
					3270	c.2155T	3270T >C									
0111	CAIS	Substitut.		4 LBD	719	2157	p.(Trp719 *)			zero			Female	Normal	pos	Sai et al. Am J Hum Genet 46: 1095, 1990
					3272	c.2157G	3272G >A									
1088	CAIS	Substitut.		4 LBD	719	2157	p.(Trp719 *)						Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
					3272	c.2157G	3272G >A									
0112	Prostate cancer	Substitut.		4 LBD	721	2161	p.(Lys721 Glu)					Somatic mutation- Bone metastases of Prostate cancer	Male	Normal		Kleinerman et al. J of Urology 155: 624A, 1996
					3276	c.2161A	3276A >G									
0113	Prostate cancer	Substitut.		4 LBD	722	2164	p.(Ala722 Thr)					Somatic mutation in 20% of isolates in initial cloning	Male	Normal		Taplin et al. New England J Med 332: 1393-1398, 1995
					3279	c.2164G	3279G >A									
0583	CAIS	Deletion		4 LBD	722	2164	p.(Ala722 fs)					1nt frameshift & stop in codon 767 - no immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
					3279	c.2164	3279 delG									
0584	CAIS	Deletion		4 LBD	722	2166	p.(Ala722 fs)					1 nt frameshift & stop - low immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
					3281	c.2166	3281 delC									
0114	CAIS	Substitut.		4 LBD	723	2169	p.(Leu723 Phe)						Female	Normal		Hiort et al: Am J Med Genet. 63: 218-222, 1996
					3284	c.2169G	3284G >T									
0761	Prostate cancer	Substitut.		4 LBD	723	2169	p.(Leu723 Phe)					patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006
					3284	c.2169G	3284G >T									
0451	CAIS	Substitut.		4 LBD	724	2170	p.(Pro724 Ser)			normal	high	-Epidiymis present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
					3285	c.2170C	3285C >T									
0791	PAIS ?	Substitut.		4 LBD	724	2171	p.(Pro724 Leu)									Mueller et al. Hum Genet 119: 681, 2006
					3286	c.2171C	3286C >T									
0452	CAIS	Substitut.		4 LBD	725	2173	p.(Gly725 Ser)			zero			Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
					3288	c.2173G	3288G >A									
0453	CAIS	Deletion/ Insertion		4 LBD	725	2174	p.(Gly725 Asp)			zero		Epididymis & Vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
					3289	c.2174_2175	3289_3290 delinsAT									
0620	CAIS	Substitut.		4 LBD	725	2174	p.(Gly725 Ala)	19		normal			Female	Normal		MacLean et al. Hum Mutat. 23: 287, 2004
					3289	c.2174G	3289G >C									
0798	CAIS	Substitut.	*	4 LBD	725	2174	p.(Gly725 Val)			zero		Severley impaired AR transactivation	Female	Normal		Jaaskelainen et al. Hum Mutat 27: 291, 2006
					3289	c.2174G	3289G >T									
0115	CAIS	Deletion		4-8 LBD						zero			Female	Normal		Brown et al. Proc Natl Acad Sci 85: 8151, 1988
1107	CAIS	Deletion		4-8 LBD									Female	Normal		
0116	CAIS	Deletion		5 LBD						zero		Affected aunt deleted for exons 6 and 7 only.	Female	Normal	pos	Maclean et al. J Clin Invest, 91: 1123, 1993
0117	CAIS	Substitut.		5 LBD						zero		p.(Tyr>Arg) substitution	Female	Normal		Marcelli et al. 74th US Endo Soc Meetings: Abstr. 224, 1992

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0118	PAIS	Substitut. 5 LBD	*	726 / 3291	2176 / c.2176T	p.(Phe726 Leu) / 3291T >C			normal	normal		Mutation disrupts N/C terminal interaction	Male	Ambiguous	pos	Quigley et al. Mech of Aging & Develop 125: 683-695, 2004
0391	PAIS	Substitut. 5 LBD		726 / 3291	2176 / c.2176T	p.(Phe726 Leu) / 3291T >C						Hypospadia and cryptorchidism	Male	Ambiguous	pos	Nordenskjold et al Urological Res, 27: 49 -55, 1999
0119	Prostate cancer	Substitut. 5 LBD	*	727 / 3295	2180 / c.2180G	p.(Arg727 Leu) / 3295G >T			normal	normal		Germ line mutation present in offspring	Male	Normal	pos	Elo et al. J Clin Endorinol Metab, 80: 3494-3500, 1995
0508	Prostate cancer	Substitut. 5 LBD	*	727 / 3295	2180 / c.2180G	p.(Arg727 Leu) / 3295G >T						Estimated that 2% of Finnish CAP patients have this mutation	Male	Normal	pos	Mononen et al. Cancer Res 60: 6479-6481, 2000
0571	Prostate Cancer	Substitut. 5 LBD		727 / 3295	2180 / c.2180G	p.(Arg727 Leu) / 3295G >T	20					Somatic mut polyGln 24 to 20 Orchiectomy Horm-refractory CaP	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
0687	Normal	Substitut. 5 LBD		727 / 3295	2180 / c.2180G	p.(Arg727 Leu) / 3295G >T						Patient suffereing frm alcoholism		Normal		Yan et al. Psychiatric Genet 14. 57-60, 2004
0120	MAIS	Substitut. 5 LBD		728 / 3299	2184 / c.2184C	p.(Asn728 Lys) / 3299C >G						Oligospermia	Male	Normal		Yong et al. Lancet, 344: 826-827, 1994
0121	PAIS	Substitut. 5 LBD		729 / 3301	2186 / c.2186T	p.(Leu729 Ser) / 3301T >C			low		*					McPhaul et al. J Clin Inv, 90: 2097, 1992
0122	Prostate Cancer	Substitut. 5 LBD	*	731 / 3306	2191 / c.2191G	p.(Val731 Met) / 3306G >A						Somatic mutation	Male	Normal		Newmark et al. Proc Natl AcadSci 89: 6319, 1992
0123	Prostate Cancer	Substitut. 5 LBD	*	731 / 3306	2191 / c.2191G	p.(Val731 Met) / 3306G >A						Somatic mutation	Male	Normal		Petersiel et al. Int J Cancer 63: 544-550, 1995
0762	Prostate Cancer	Substitut. 5 LBD	*	731 / 3306	2191 / c.2191G	p.(Val731 Met) / 3306G >A						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006
0125	CAIS	Substitut. 5 LBD	*	733 / 3312	2197 / c.2197G	p.(Asp733 Tyr) / 3312G >T				high			Female	Normal		Brown et al. 74th US Endo Soc Meeting, Abstr 1506, 1992
0126	CAIS	Substitut. 5 LBD		733 / 3312	2197 / c.2197G	p.(Asp733 Tyr) / 3312G >T			zero				Female	Normal		Pinsky et al. Clin Inv Med 15: 456, 1992
0127	CAIS	Substitut. 5 LBD		733 / 3312	2197 / c.2197G	p.(Asp733 Tyr) / 3312G >T							Female	Normal		Ghirri and Brown. Pediatr Res 33: Abstr 95, 1993
0801	CAIS	Substitut. 5 LBD		733 / 3312	2197 / c.2197G	p.(Asp733 Tyr) / 3312G >T						6 affected family members	Female	Normal	pos	Scott et al. Endocr Pract 12: 664-669, 2006
0124	CAIS	Substitut. 5 LBD		733 / 3312	2197 / c.2197G	p.(Asp733 Asn) / 3312G >A				high			Female	Normal		Brown et al. 74th US Endo Soc Meeting, Abstr 1506, 1992
0659	CAIS	Substitut. 5 LBD		733 / 3312	2197 / c.2197G	p.(Asp733 Asn) / 3312G >A						Epididymis & Vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0310	CAIS	Substitut. 5 LBD		733 / 3312	2197 / c.2197G	p.(Asp733 Asn) / 3312G >A	19						Female	Normal		Ko et al. J Reprod. Med 42: 424- 427, 1997
0955	CAIS	Substitut. 5 LBD		733 / 3312	2197 / c.2197G	p.(Asp733 Asn) / 3312G >A							Female	Normal	neg	Wu et al. Fertility & Sterility 93: 2076, e1-4, 2010
0628	Prostate cancer	Substitut. 5 LBD		733 / 3314	2199 / c.2199C	p.(Asp733 Asp) / 3314C >T						Orch + Bicalutiamide +phosphamide treat Gleason 10. Somatic	Male	Normal		Haapala et al. Lab Invest. 81: 1647-1651, 2001
0752	CAIS	Substitut. 5 LBD		734 / 3315	2200 / c.2200C	p.(Gln734 *) / 3315C >T						No WD development	Female	Normal	neg	Barbaro et al. Clin Endocrinol 66: 822 -826, 2007
0128	PAIS	Substitut. 5 LBD		734 / 3317	2202 / c.2202G	p.(Gln734 His) / 3317G >T						This patient was a mosaic for wt. & mut. alleles- de novo mut.	Female	Ambiguous	neg	Hiort et al. J Pediatrics 132: 939- 943, 1998
0726	CAIS	Substitut. 5 LBD		738 / 3327	2212 / c.2212A	p.(Ile738 Phe) / 3327A >T							Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
0129	PAIS	Substitut. 5 LBD	*	738 / 3328	2213 / c.2213T	p.(Ile738 Thr) / 3328T >C			low			Mutation disrupts N/C terminal interaction	Male	Ambiguous	pos	Quigley et al. Mech Aging & Develop 125: 683-695
0944	CAIS	Substitut. 5 LBD	*	739 / 3331	2216 / c.2216A	p.(Gln739 Arg) / 3331A >G						V reduced transactiv reduced interaction with FxxLX	Female	Normal		Elfferich et al. Sexual Development 3: 237 -244, 2009

Accession # / Phenotype	Mutation type	Exon / Domain	Pathogenicity proven	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0530	CAIS	Substitut.	5 LBD	*	740 3333	2218 c.2218T	p.(Tyr740 Asp) 3333T >G			zero			no transactivation in COS-1 cells	Female	Normal		Suzuki et al. Int. J Andrology 24: 183-188, 2001
0616	PAIS	Substitut.	5 LBD	*	741 3337	2222 c.2222C	p.(Ser741 Cys) 3337C >G			v low			Acute stress masked PAIS.	Male	Ambiguous	pos	Pitteloud et al. J Clin Endorinol & Metab 89: 1053-1058, 2004
0130	CAIS	Substitut.	5 LBD	*	742 3339	2224 c.2224T	p.(Trp742 Arg) 3339T >C			low				Female	Normal	neg	Marcelli et al. J Clin Invest 94: 1642-1650, 1994
0360	Prostate cancer	Substitut.	5 LBD		742 3340	2225 c.2225G	p.(Trp742 *) 3340G >A						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
0595	CAIS	Substitut.	5 LBD	*	742 3340	2225 c.2225G	p.(Trp742 Leu) 3340G >T						Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
0858	CAIS	Substitut.	5 LBD		742 3340	2225 c.2225G	p.(Trp742 Leu) 3340G >T						Diag at birth bilateral gonadectomy at 14yr	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0859	CAIS	Substitut.	5 LBD		742 3340	2225 c.2225G	p.(Trp742 Leu) 3340G >T						Diag at birth bilateral gonadectomy at 12.5yr	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0629	Prostate cancer	Substitut.	5 LBD		742 3341	2226 c.2226G	p.(Trp742 Cys) 3341G >T						Orch + bicalutamide Gleason 10- somatic mutation	Male	Normal		Haaplaa et al. Lab Invest. 81: 1647-1651, 2001
0635	PAIS	Substitut.	5 LBD		742 3341	2226 c.2226G	p.(Trp742 Cys) 3341G >C	27		low	high		Testes located in scrotum - Also has p.(Leu548Phe mutation	Male	Ambiguous	pos	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
0552	Prostate cancer	Substitut.	5 LBD		742 3341	2226 c.2226G	p.(Trp742 Cys) 3341G >T						Treated with bicalutamide - somatic mutation	Male	Normal		Taplin et al. J Clinical Oncology 21: 2673 -2678, 2003
0982	CAIS	Substitut.	5 LBD		742 3339	2224 c.2224T	p.(Trp742 Arg) 3339T >C	23	17					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0131	PAIS	Substitut.	5 LBD		743 3342	2227 c.2227A	p.(Met743 Val) 3342A >G					high					Ris-Stalpers et al. Pediatric Res. 36: 227 -234,1994
0341	PAIS	Substitut.	5 LBD		743 3342	2227 c.2227A	p.(Met743 Val) 3342A >G	21		zero			Testis located in inginual region	Female	Ambiguous	pos	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
0744	PAIS	Substitut.	5 LBD		743 3342	2227 c.2227A	p.(Met743 Val) 3342A >G						affected neice of 0131	Female	Ambiguous		Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0132	PAIS	Substitut.	5 LBD	*	743 3344	2229 c.2229G	p.(Met743 Ile) 3344G >A			normal	high		*	Female	Ambiguous		Batch et al. Hum Mol Genet 1: 497, 1992
0948	PAIS	Substitut.	5 LBD		743 3344	2229 c.2229G	p.(Met743 Ile) 3344G >A							Female	Ambiguous		Nagaraja et al. J Pediatr Endocrinol Metab 22: 1169-1173, 2009
1006	PAIS	Substitut.	5 LBD		743 3344	2229 c.2229G	p.(Met743 Ile) 3344G >A	18	18	low	normal		Mother heterozygote carrier	Male	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0133	PAIS	Substitut.	5 LBD	*	744 3346	2229 c.2231G	p.(Gly744 Val) 3346G >T			low		high	Transcription only at high conc of androgen	Female	Ambiguous		Georget et al. J Clin Endocrinol Metab 83: 3597-3603, 1998
0134	PAIS	Substitut.	5 LBD		744 3346	2229 c.2231G	p.(Gly744 Val) 3346G >T			normal	normal		*				Nakao et al. J Clin Endocrinol Metab 77: 103-107, 1993
0414	CAIS	Substitut.	5 LBD		744 3346	2229 c.2231G	p.(Gly744 Val) 3346G >T			zero			de novo mutation	Female	Normal		Lobaccaro et al. J Steroid Biochem & Mol Biol. 44: 211-216, 1993
0536	CAIS	Substitut.	5 LBD		744 3346	2229 c.2231G	p.(Gly744 Glu) 3346G >A			normal				Female	Normal		Chavez et al. J Hum Genet. 46: 560-56, 2001
0564	CAIS	Substitut.	5 LBD	*	744 3346	2229 c.2231G	p.(Gly744 Glu) 3346G >A			v low	high	high	*	Female	Normal		Pujol et al. J Clin Endorinol & Metab. 87: 5793-5800. 2002
0572	Prostate cancer	Substitut.	5 LBD		744 3347	2232 c.2232G	p.(Gly744) 3347G >C						Somatic mutation + estrogen treatment Horm-refractory CaP	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
0361	Prostate cancer	Deletion	5 LBD		744 3347	2232 c.2232	p.(Gly744 fs) 3347 delG						Frameshift-somatic mut.- separate tumor in same indv. as 0362	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
0519	CAIS	Substitut.	5 LBD		744 3345	2230 c.2230G	p.(Gly744 Arg) 3345G >C							Female	Normal		Chavez et al. Clin Genet 59: : 185-188, 2001

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base / HGVS nt / HGVS base	Change: Amino acid Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0983 CAIS	Substitut.	5 LBD		744 2231 3346 c.2231G	p.(Gly744 Val) 3346G >T	24	17					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0135 CAIS	Substitut.	5 LBD		745 2233 3348 c.2233C	p.(Leu745 Phe) 3348C >T						1 affected sibling			pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0984 CAIS	Substitut.	5 LBD		745 2233 3348 c.2233C	p.(Leu745 Phe) 3348C >T	21	17				1 heterozygote sister & 1 wt sister	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0362 Prostate cancer	Substitut.	5 LBD		745 2233 3348 c.2237T	p.(Leu745 Phe) 3348C >T						Somatic mutation - separate tumor in same indv. as 0361	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
0136 PAIS	Substitut.	5 LBD		746 2237 3352 c.2237T	p.(Met746 Thr) 3352T >C			zero							Ris-Stalpers et al. Pediatric Res 36: 227 -234, 1994
0727 CAIS	Substitut.	5 LBD		746 2237 3352 c.2237T	p.(Met746 Thr) 3352T >C						Mother and sisters heterozygotous	Female	Normal	pos	Ledig et al: Horm Res 63: 263-269, 2005
0755 CAIS	Substitut.	5 LBD	*	746 2238 3353 c.2238G	p.(Met746 Ile) 3353G >C						Enanced affinity for & increased transact for estradiol	Female	Normal		Bonagura et al. Mol Cell Endocrinol 263: 79 -89, 2007
1007 PAIS	Substitut.	5 LBD		746 2236 3351 c.2236A	p.(Met746 Leu) 3351A >C	23	17				mother & grandmother heterozyote carriers	Male	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0138 PAIS	Substitut.	5 LBD		747 2239 3354 c.2239G	p.(Val747 Met) 3354G >A							Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0137 PAIS	Substitut.	5 LBD		747 2239 3354 c.2239G	p.(Val747 Met) 3354G >A										Brown et al. 74th US Endo Soc Meeting, Abstr 1506, 1992
0949 PAIS	Substitut.	5 LBD		747 2239 3354 c.2239G	p.(Val747 Met) 3354G >A										Nagaraja et al. J Pediatr Endocrinol Metab 22: 1169-1173, 2009
0785 MAIS	Substitut.	5 LBD		748 2242 3357 c.2242T	p.(Phe748 Ile) 3357T >A						Male Infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
0985 CAIS	Substitut.	5 LBD		748 2243 3358 c.2243T	p.(Phe748 Cys) 3358T >G	23	17				1 wt sister	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0763 Prostate cancer	Deletion	5 LBD		748 2244 3359 c.2244	p.(Phe748 fs) 3359 delT						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006
0492 Prostate cancer	Substitut.	5 LBD		749 2245 3360 c.2245G	p.(Ala749 Thr) 3360G >A						Also p.(S866P; Q868* and Q920R). Somatic mutations	Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000
0565 Prostate cancer	Substitut.	5 LBD	*	749 2245 3360 c.2245G	p.(Ala749 Thr) 3360G >A			normal		high	Somatic mutation. weaker interaction with HSP	Male	Normal		James et al. Mol Endocrinol. 16: 2692 -2705, 2002
0139 PAIS	Substitut.	5 LBD	*	749 2246 3361 c.2246C	p.(Ala749 Asp) 3361C >A			low		high	Abnormal dissociation				Marcelli et al. J Clin Invest 94: 1642-1650, 1994
0363 Prostate cancer	Substitut.	5 LBD		749 2246 3361 c.2246C	p.(Ala749 Val) 3361C >T						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
0140 CAIS	Substitut.	5 LBD		750 2248 3363 c.2248A	p.(Met750 Val) 3363A >G							Female	Normal	pos	DeBellis et al. Mol Endocrinol 6: 1909-20, 1992
0141 CAIS	Substitut.	5 LBD		750 2248 3363 c.2248A	p.(Met750 Val) 3363A >G							Female	Normal	pos	Jakubiczka et al. Hum Genet 90: 311-2, 1992
0483 PAIS	Substitut.	5 LBD		750 2248 3363 c.2248A	p.(Met750 Val 3363A >G)			normal	high						Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0614 CAIS	Substitut.	5 LBD		750 2248 3363 c.2248A	p.(Met750 Val) 3363A >G						Fallopian tube present	Female	Normal		Van et al. Eur J. Pediatr. 162: 781-784, 2003
0679 CAIS	Substitut.	5 LBD		750 2248 3363 c.2248A	p.(Met750 Val) 3363A >G						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0680 CAIS	Substitut.	5 LBD		750 2248 3363 c.2248A	p.(Met750 Val) 3363A >G						Diff between ext genital & chrom - in amniocentesis	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0364 Prostate cancer	Substitut.	5 LBD		750 2250 3365 c.2250G	p.(Met750 Ile) 3365G >A						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0630	Prostate cancer	Substitut.	5 LBD	750	2250	p.(Met750 Ile)						Orch + bicalutamide Gleason 10 +silent mut Q868Q(G>A)	Male	Normal		Haaplaa et al. Lab Invest. 81: 1647-1651, 2001
				3365	c.2250G	3365G >A										
0365	Prostate cancer	Substitut.	5 LBD	751	2251	p.(Gly751 Ser)						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
				3366	c.2251G	3366G >A										
0142	CAIS	Substitut.	5 LBD	* 751	2252	p.(Gly751 Asp)			v low			Mutation found in two unrelated patients	Female	Normal		Bevan et al. J Steroid Biochem Mole Biol 61: 19-26, 1997
				3367	c.2252G	3367G >A										
0143	CAIS	Substitut.	5 LBD	751	2252	p.(Gly751 Asp)							Female	Normal		Brown et al: 74th US Endo Soc Meeting Abstr 1506, 1992
				3367	c.2252G	3367G >A										
0144	CAIS	Substitut.	5 LBD	752	2254	p.(Trp752 Arg)			low	high		Sibling of 0703	Female	Normal	pos	Brinkmann et al. J Steroid Biochem Mol Biol 53: 443, 1995
				3369	c.2254T	3369T >A										
0366	Prostate cancer	Substitut.	5 LBD	752	2255	p.(Trp752 *)						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
				3370	c.2255G	3370G >A										
0367	Prostate cancer	Substitut.	5 LBD	752	2255	p.(Trp752 *)						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
				3370	c.2255G	3370G >A										
0368	Prostate cancer	Substitut.	5 LBD	752	2256	p.(Trp752 *)						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
				3371	c.2256G	3371G >A										
0401	CAIS	Substitut.	5 LBD	752	2256	p.(Trp752 *)			zero				Female	Normal		Yaegashi et al. Tohoku J of Exp Med 187: 263 -272, 1999
				3371	c.2256G	3371G >A										
0664	PAIS	Substitut.	5 LBD	752	2256	p.(Trp752 *)						Somatic mosaicism - male changed to female at 18mths	Female	Ambiguous		Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
				3371	c.2256G	3371G >A										
0703	CAIS	Substitut.	5 LBD	752	2254	p.(Trp752 Arg)			normal	high		Sibling of 0144	Female	Normal		Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
				3369	c.2254T	3369T >A										
1089	CAIS	Substitut.	5 LBD	* 753	2257	p.(Arg753 *)							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
				3372	c.2257C	3372C >T										
0145	CAIS	Substitut.	5 LBD	* 753	2257	p.(Arg753 *)			zero				Female	Normal		Pinsky et al. Clin Inv Med 15: 456, 1992
				3372	c.2257C	3372C >T										
0146	CAIS	Substitut.	5 LBD	* 753	2257	p.(Arg753 *)							Female	Normal		Brinkmann et al. J Steroid Biochem Mol Biol 53: 443, 1995
				3372	c.2257C	3372C >T										
0342	CAIS	Substitut.	5 LBD	* 753	2257	p.(Arg753 *)	16		zero			Testes located in abdomen	Female	Normal	pos	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
				3372	c.2257C	3372C >T										
0402	CAIS	Substitut.	5 LBD	* 753	2257	p.(Arg753 *)			zero				Female	Normal		Yaegashi et al. Tohoku J of Exp Med 187: 263 -272, 1999
				3372	c.2257C	3372C >T										
0728	CAIS	Substitut.	5 LBD	* 753	2257	p.(Arg753 *)							Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
				3372	c.2257C	3372C >T										
0754	CAIS	Substitut.	5 LBD	* 753	2258	p.(Arg753 Gln)						Inherted from maternal grandmother present in two sisters	Female	Normal	pos	Olafsson et al. Laeknabladid 86: 263 -166, 2000
				3373	c.2258G	3373G >A										
0147	CAIS	Substitut.	5 LBD	* 753	2258	p.(Arg753 Gln)			zero			Mutation found in two unrel. families. Equivalent to tfm rat	Female	Normal		Brown et al. 74th US Endo Soc Meeting, Abstr 1506, 1992
				3373	c.2258G	3373G >A										
0333	CAIS	Substitut.	5 LBD	* 753	2258	p.(Arg753 Gln)							Female	Normal	pos	Komori et al. Arch Gynecol & Obstetrics 261: 95-100, 1998
				3373	c.2258G	3373G >A										
0349	CAIS	Substitut.	5 LBD	* 753	2258	p.(Arg753 Gln)							Female	Normal		Cabral et al. Brazilian J Med & Biol Res. 31: 775-758, 1998
				3373	c.2258G	3373G >A										
0148	CAIS	Substitut.	5 LBD	* 753	2258	p.(Arg753 Gln)			zero			Equivalent to tfm rat	Female	Normal		Evans. J Endocrinol 135 Suppl, Abstr P26, 1992
				3373	c.2258G	3373G >A										
0497	CAIS	Substitut.	5 LBD	* 753	2258	p.(Arg753 Gln)						Bilateral testicular tumors	Female	Normal		Sakai et al. IntJ of Urology 7: 390-392, 2000
				3373	c.2258G	3373G >A										
0860	CAIS	Substitut.	5 LBD	* 753	2258	p.(Arg753 Gln)						Prenatal diagnosis	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
				3373	c.2258G	3373G >A										
1090	CAIS	Substitut.	5 LBD	* 753	2258	p.(Arg753 Gln)							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
				3373	c.2258G	3373G >A										

Accession #	Phenotype	Mutation type	Pathogenicity proven Exon	Domain	CpG hot spot	Position Amino acid	Base	HGVS nt	HGVS base	Change Amino acid	Base	Exon 1 tracts Poly Gln #	Poly Gly #	Androgen Binding Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
1091	CAIS	Substitut.	5	LBD	*	753	3373	2258	c.2258G	p.(Arg753 Pro)	3373G >C								Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0149	CAIS	Substitut.	5	LBD		755	3378	2263	c.2263T	p.(Phe755 Val)	3378T >G			zero					Female	Normal		Lobaccaro et al. Hum Mol Genet 2: 1041 -1043, 1993
0369	Prostate cancer	Substitut.	5	LBD		755	3378	2263	c.2263T	p.(Phe755 Leu)	3378T >C							Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
0879	CAIS	Substitut.	5	LBD	*	755	3378	2263	c.2263T	p.(Phe755 Val)	3378T >G			high				Greatly reduced transactivation 5%	Female	Normal		Tadokoro et al. Clinical Endocrinology 71: 253 -260, 2009
0150	CAIS	Substitut.	5	LBD		755	3378	2263	c.2263T	p.(Phe755 Val)	3378T >G								Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0880	PAIS	Substitut.	5	LBD	*	755	3379	2264	c.2264T	p.(Phe755 Ser)	3379T >C			normal				Microphallus no hypospadias	Male	Ambiguous		Tadokoro et al. Clinical Endocrinology 71: 253 -260, 2009
1050	MAIS	Substitut.	5	LBD	*	755	3379	2264	c.2264T	p.(Phe755 Ser)	3379T >C							Azoospermia. Fertile - intracytoplasmic sperm injection	Male	Normal		Massin et al. Clin Endorinol (Oxf) 77:593 -598, 2012
0715	PAIS	Substitut.	5	LBD	*	755	3380	2265	c.2265C	p.(Phe755 Leu)	3380C >A			high				Microphallus, hypospadias, cryptorchisms	Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
0152	PAIS	Substitut.	5	LBD	*	755	3380	2265	c.2265C	p.(Phe755 Leu)	3380C >A			normal	high		*		Male	Ambiguous		Weidemann et al. Clin Endocrinology 45: 733 -739, 1996
0151	PAIS	Substitut.	5	LBD		755	3380	2265	c.2265C	p.(Phe755 Leu)	3380C >A								Male	Ambiguous		Hiort et al: Hum Mol Genet 3: 1163-1166 1994
0714	PAIS	Substitut.	5	LBD		755	3380	2265	c.2265C	p.(Phe755 Leu)	3380C >A			high			*		Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
0370	Prostate cancer	Substitut.	5	LBD		756	3381	2266	c.2266A	p.(Thr756 Ala)	3381A >G							Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
0602	Prostate cancer	Substitut.	5	LBD		757	3384	2269	c.2269A	p.(Asn757 Asp)	3384A >G							Flutamide treated - somatic mutation	Male	Normal		Taplin et al. J Clinical Oncology 21: 2673 -2678, 2003
0153	PAIS	Substitut.	5	LBD		757	3385	2270	c.2270A	p.(Asn757 Ser)	3385A >G								Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0532	MAIS	Substitut.	5	LBD	*	757	3385	2270	c.2270A	p.(Asn757 Ser)	3385A >G			high				Severe oligospermia- transactivation 38% of wt.	Male	Normal		Giwercman et al. Clin Endocrinol 54: 827 -834, 2001
1039	PAIS	Substitut.	5	LBD		757	3385	2270	c.2270A	p.(Asn757 Ser)	3385A >G							External Masculisation Score = 12	Male	Ambiguous	pos	Hellmann et al. Arch Dis Child 97:403-409, 2012
1040	PAIS	Substitut.	5	LBD		757	3385	2270	c.2270A	p.(Asn757 Ser)	3385A >G							External Masculisation Score = 12	Male	Ambiguous	pos	Hellmann et al. Arch Dis Child 97:403-409, 2012
0573	Prostate cancer	Substitut.	5	LBD		758	3387	2272	c.2272G	p.(Val758 Ile)	3387G >A							Somatic mutation + Orchiectomy Horm-refractor y CaP	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
0300	Prostate cancer	Substitut.	5	LBD	*	758	3388	2273	c.2273T	p.(Val758 Ala)	3388T >C							Binds R1881 norm.- transcriptionally inactive- Som mut	Male	Normal		James et al. 79th US Endo Soc Meeting, Abstr P2-484, 1997
0493	Prostate cancer	Substitut.	5	LBD		758	3388	2273	c.2273T	p.(Val758 Ala)	3388T >C							Somatic mutation	Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000
0346	PAIS	Substitut.	5	LBD	*	759	3391	2276	c.2276A	p.(Asn759 Thr)	3391A >C			normal		high	*	50% reduction in transactivation in COS-7				Yong et al. Mol & Cell Endocrinol. 137: 41-50, 1998
0371	Prostate cancer	Substitut.	5	LBD		760	3393	2278	c.2278T	p.(Ser760 Pro)	3393T >C							Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
0154	CAIS	Substitut.	5	LBD		760	3394	2279	c.2279C	p.(Ser760 Phe)	3394C >T			zero					Female	Normal		DeBellis et al. Mol Endocrinol, 6: 1909-20, 1992
1092	CAIS	Substitut.	5	LBD		760	3394	2279	c.2279C	p.(Ser760 Tyr)	3394C >A								Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0605	CAIS	Deletion/ insertion	5	LBD		761				p.(Arg761 fs)				zero				also 3396_3402del7 & 3408_3418ins11 stop 9 codon downstr.	Female	Normal	pos	Vichlis et al. J Hum Genet 48: 346-351, 2003

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Androgen Binding: Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0841	PAIS	Substitut.		761		p.(Arg761 Ser)										Jeske et al. J Pediatr Endocrinol Metab 20: 893-908, 2007
0938	Prostate cancer	Substitut.		761		p.(Arg761)						Silent mut. Treated with anti-androgens. Occurred in >2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0729	PAIS	Substitut.		762	2285	p.(Met762 Thr)						mother heterozygous	Male	Ambiguous	pos	Ledig et al: Horm Res 63: 263-269, 2005
		5 LBD		3400	c.2285T	3400T >C										
0155	CAIS	Substitut.		763	2287	p.(Leu763 Phe)			zero				Female	Normal		Brown et al. 74th US Endo Soc Meeting, Abstr 1506, 1992
		5 LBD		3402	c.2287C	3402C >T										
0156	CAIS	Substitut.	*	763	2287	p.(Leu763 Phe)			zero				Female	Normal		Bevan et al. J Steroid Biochem Mole Biol 61: 19-26, 1997
		5 LBD		3402	c.2287C	3402C >T										
0861	CAIS	Substitut.		763	2287	p.(Leu763 Phe)						Diag at 7yr bilateral gonadectomy at 8yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
		5 LBD		3402	c.2287C	3402C >T										
0862	CAIS	Substitut.		763	2287	p.(Leu763 Phe)						Diagnosed at 1 wk	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
		5 LBD		3402	c.2287C	3402C >T										
0157	CAIS	Substitut.		764	2290	p.(Tyr764 His)							Female	Normal		Quigley et al. Endocrin. Reviews, 16: 271, 1995
		5 LBD		3405	c.2290T	3405T >C										
0159	PAIS	Substitut.		764	2291	p.(Tyr764 Cys)			low				Male	Ambiguous		Morono et al. Human Mutation 6: 152-162, 1995
		5 LBD		3406	c.2291A	3406A >G										
0405	PAIS	Substitut.		764	2291	p.(Tyr764 Cys)							Male	Ambiguous		Batch et al. Arch Disease Child 68: 453, 1993
		5 LBD		3406	c.2291A	3406A >G										
0484	PAIS	Substitut.		764	2291	p.(Tyr764 Cys)			normal	high						Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		5 LBD		3406	c.2291A	3406A >G										
0485	PAIS	Substitut.		764	2291	p.(Tyr764 Cys)			normal	high						Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		5 LBD		3406	c.2291A	3406A >G										
0636	PAIS	Substitut.		764	2291	p.(Tyr764 Cys)	22		low			Gynocomastia -testis located in scrotum	Male	Ambiguous	neg	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
		5 LBD		3406	c.2291A	3406A >G										
0372	Prostate Cancer	Substitut.		764	2291	p.(Tyr764 Cys)						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
		5 LBD		3406	c.2291A	3406A >G										
0158	PAIS	Substitut.	*	764	2291	p.(Tyr764 Cys)	12		normal	high	*	PolyGln tract short (only 12 repeats)	Male	Ambiguous	pos	McPhaul et al.J Clin Inv 87: 1413,1991: Batch&al Arc Dis Ch
		5 LBD		3406	c.2291A	3406A >G										
0850	CAIS	Substitut.		764		p.(Tyr764 *)						Diag at 1 wk-Bilateral gonadectomy at 3wks	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
		5 LBD														
0160	CAIS	Substitut.	*	765		p.(Phe765 Leu)			low		high		Female	Normal	neg	Marcelli et al. J Clin Invest 94: 1642-1650, 1994
		5 LBD														
0161	CAIS	Substitut.		765	2293	p.(Phe765 Leu)			zero				Female	Normal		Ris-Stalpers et al: Pediatric Res,36. 227 -234, 1994
		5 LBD		3408	c.2293T	3408T >C										
0162	CAIS	Substitut.		765	2295	p.(Phe765 Leu)			low	normal			Female	Normal		Pinsky et al. Clin Inv Med, 15: 456, 1992
		5 LBD		3410	c.2295C	3410C >G										
0163	CAIS	Substitut.	**	766	2296	p.(Ala766 Thr)			zero				Female	Normal		Bevan et al. J Steroid Biochem Mole Biol 61: 19-26, 1997
		5 LBD		3411	c.2296G	3411G >A										
0164	CAIS	Substitut.	*	766	2296	p.(Ala766 Thr)			zero				Female	Normal		Merkabi et al.75th US Endo Soc Meeting Abstr 602, 1993
		5 LBD		3411	c.2296G	3411G >A										
0165	CAIS	Substitut.	*	766	2296	p.(Ala766 Thr)							Female	Normal		Sweet et al. Fertil Sterilty 58: 703, 1992
		5 LBD		3411	c.2296G	3411G >A										
0166	CAIS	Substitut.	*	766	2296	p.(Ala766 Thr)			zero				Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996
		5 LBD		3411	c.2296G	3411G >A										
0311	CAIS	Substitut.	*	766	2296	p.(Ala766 Thr)	27						Female	Normal		Ko et al. J Reprod. Med 42: 424- 427, 1997
		5 LBD		3411	c.2296G	3411G >A										
0382	CAIS	Substitut.	*	766	2296	p.(Ala766 Thr)							Female	Normal		Giwercman et al. Human Genetics 103: 529-531, 1998
		5 LBD		3411	c.2296G	3411G >A										

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0454	CAIS	Substitut.	*	766	2296	p.(Ala766 Thr)							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		5 LBD		3411	c.2296G	3411G >A										
0455	CAIS	Substitut.	*	766	2296	p.(Ala766 Thr)							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		5 LBD		3411	c.2296G	3411G >A										
0456	CAIS	Substitut.	*	766	2296	p.(Ala766 Thr)						Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		5 LBD		3411	c.2296G	3411G >A										
0585	CAIS	Substitut.		766	2296	p.(Ala766 Thr)						Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
		5 LBD		3411	c.2296G	3411G >A										
0586	PAIS	Substitut.		766	2296	p.(Ala766 Thr)						Reduced immunoreactive AR	Female	Ambiguous		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
		5 LBD		3411	c.2296G	3411G >A										
0730	CAIS	Substitut.		766	2296	p.(Ala766 Thr)							Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
		5 LBD		3411	c.2296G	3411G >A										
0520	PAIS	Substitut.		766	2296	p.(Ala766 Ser)										Chavez et al. Clin Genet 59: 185-188, 2001
		5 LBD		3411	c.2296G	3411G >T										
0167	CAIS	Substitut.		766	2297	p.(Ala766 Val)	20		zero				Female	Normal		Pinsky et al, Clin Inv Med, 15: 456-472, 1992
		5 LBD		3412	c.2297C	3412C >T										
0168	CAIS	Substitut.	*	767	2299	p.(Pro767 Ser)			low	high	high		Female	Normal	pos	Marcelli et al. J Clin Invest 94: 1642-1650. 1994
		5 LBD		3414	c.2299C	3414C >T										
0457	CAIS	Substitut.		767	2299	p.(Pro767 Ser)							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		5 LBD		3414	c.2299C	3414C >T										
0681	CAIS	Substitut.		767	2299	p.(Pro767 Ala)						bilateral inguinal hernia	Female	Normal		Bouvattier et al. J Clin Endocrinol Metab 87: 29-32, 2002
		5 LBD		3414	c.2299C	3414C >G										
0543	CAIS	Substitut.		767	2299	p.(Pro767 Ala)			normal	high		2 affcted siblings variable phenotype	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
		5 LBD		3414	c.2299C	3414C >G										
0611	CAIS	Substitut.		767	2299	p.(Pro767 Ala)						Twin sisters	Female	Normal	pos	Correa et al. 16th Meet. Latin Amer.Soc Ped Endo. Abstr: 26, 2003
		5 LBD		3414	c.2299C	3414C >G										
0587	CAIS	Deletion		767	2300	p.(Pro767 fs)						1 nt del.-frameshift & stop, low immunoreact AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
		5 LBD		3415	c.2300	3415 delC										
0169	CAIS	Deletion		767	2301	p.(Pro767 fs)						Single nt deletion causing frameshift & stop in codon 808	Female	Normal	pos	Baldazzi et al. Hum Mol Genet 3: 1169-1170, 1994
		5 LBD		3416	c.2301	3416 delT										
0388	CAIS	Deletion		767	2301	p.(Pro767 fs)						Single nt deletion causing frameshift & stop in codon 808	Female	Normal		Chung et al. Molecules & Cells 8: 741-745, 1998
		5 LBD		3416	c.2301	3416 delT										
0458	CAIS	Deletion		767	2301	p.(Pro767 fs)						1nt del-frameshit & stop in codon 808 Wolffian remnants	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		5 LBD		3416	c.2301	3416 delT										
0459	CAIS	Deletion		767	2301	p.(Pro767 fs)						1nt del-frameshit & stop in codon 808 Wolffian remnants	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		5 LBD		3416	c.2301	3416 delT										
0561	CAIS	Deletion		767	2301	p.(Pro767 fs)						1nt del frame shift & stop in codon 808 in 2 unrelat individs	Female	Normal		Soriano Guillén et al. An Esp Pediatr 56: 347 -352, 2002
		5 LBD		3416	c.2301	3416 delT										
0588	CAIS	Deletion		767	2301	p.(Pro767 fs)						1 nt del framshift & stop codon 808 no immunoreact AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
		5 LBD		3416	c.2301	3416 delT										
0853	CAIS	Deletion		767	2301	p.(Pro767 fs)						1nt del framshift & stop in codon 808 Diag - 7yr Wolff rem	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
		5 LBD		3416	c.2301	3416 delT										
0953	CAIS	Deletion		767	2301	p.(Pro767 fs)						1nt del framshift & stop in codon 808 Male gender identity	Female	Normal		T'Sjoen et al. Arch Sex Behav 40: 655-638, 2011
		5 LBD		3416	c.2301	3416 delT										
0665	PAIS	Substitut.		767	2299	p.(Pro767 Ser)						Somatic mosaicism	Female	Ambiguous		Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
		5 LBD		3414	c.2299C	3414C >T										
0170	CAIS	Substitut.		768	2304	p.(Asp768 Glu)			v low				Female	Normal		Lobaccaro et al. Pediatr Res 33: Abstr. 115, 1993
		5 LBD		3419	c.2304T	3419T >G										
0343	CAIS	Substitut.		768	2304	p.(Asp768 Glu)							Female	Normal		Melo et al. 80th US Endo Soc Meeting Abstr P2-44, 1998
		5 LBD		3419	c.2304T	3419T >G										

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base / HGVS nt / HGVS base	Change: Amino acid Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0893 CAIS	Substitut.	5 LBD		768 2302 3417 c.2302G	p.(Asp768 Tyr) 3417G >T						Bone size intermediate between male and female	Female	Normal		Taes et al. Bone 45: 392-397, 2009
0544 PAIS	Substitut.	5 LBD		769 2305 3420 c.2305C	p.(Leu 769 Met) 3420C >A			normal	high		2 affected siblings with variable phenotype	Female	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0637 CAIS	Substitut.	5 LBD		769 2305 3420 c.2305C	p.(Leu769 Val) 3420C >G	20					Testis located in labia majora	Female	Normal		Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
0460 CAIS	Substitut.	5 LBD		769 2306 3421 c.2306T	p.(Leu769 Pro) 3421T >C							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0666 CAIS	Deletion/ Insertion	5 LBD		771 2312 3427 c.2312_2313	p.(Phe771 *) 3427_3428 delinsAA						Somatic mosaicism sister heterozygous mother som mosaic ?	Female	Normal	pos	Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
0171 PAIS	Substitut.	5 LBD		772 2314 3429 c.2314A	p.(Asn772 His) 3429A >C							Female	Ambiguous		Hiort et al. Hum Mol Genet 3: 1163-1166 1994
0526 PAIS	Substitut.	5 LBD	*	772 2314 3429 c.2314A	p.(Asn772 His) 3429A >C				high		Size & level of expression of AR normal	Female	Ambiguous		Zhu et al, 83rd US Endo Soc Meeting, Abstr P2-34, 2001
0173 PAIS	Substitut.	5 LBD		773 2318 3433 c.2318A	p.(Glu773 Gly) 3433A >G			low	high						Tincello et al. Clinical Endocrinology 46: 497 -506, 1997
0174 PAIS	Substitut.	5 LBD	*	773 2318 3433 c.2318A	p.(Glu773 Ala) 3433A >C	25	23	normal	normal	high		Male	Ambiguous		Shkolny et al. J Clin Endocrinol Metab 84: 805-810, 1999
0172 CAIS	Substitut.	5 LBD		773 2317 3432 c.2317G	p.(Glu773 *) 3432G >T			zero				Female	Normal		Imasaki et al. Endocrine Journal 42: 643-648 1995
0839 CAIS	Deletion	5 LBD		773 2317 3432 c.2317_2320	p.(Glu773 fs) 3432_3435 delGATG						4 nt deletion leading to stop in codon 788	Female	Normal		Jeske et al. J Pediatr Endocrinol Metab 20: 893-908, 2007
0336 CAIS	Substitut.	6 LBD	**	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T	26	23	normal	normal			Female	Normal		Prior et al. Am J Hum Genet 51: 143, 1992
0176 CAIS	Substitut.	6 LBD	**	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T	27	19	zero				Female	Normal	pos	Prior et al. Am J Hum Genet 51: 143, 1992
0177 CAIS	Substitut.	6 LBD	*	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T			zero				Female	Normal		Mebarki et al. 72nd US Endo Soc Meeting, Abstr 791, 1990
0178 CAIS	Substitut.	6 LBD	*	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T							Female	Normal		Hiort et al. J Pediatrics 132: 939-943, 1998
0179 CAIS	Substitut.	6 LBD	*	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T			v low	high			Female	Normal	neg	Marcelli et al. J Clin Endocrinol Metab 73: 318, 1991
0180 CAIS	Substitut.	6 LBD	*	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T							Female	Normal		Jakubiczka et al. Human Mutation 9: 57 -61, 1997
0331 CAIS	Substitut.	6 LBD	*	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T							Female	Normal		Komori et al. Arch Gynecol & Obstetrics 261: 95-100, 1998
0175 CAIS	Substitut.	6 LBD	**	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T			v low				Female	Normal		Brown et al. Mol Endocrinol, 4: 1759-72, 1990
0355 CAIS	Substitut.	6 LBD	*	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T						mosaic-de novo mutation	Female	Normal	neg	Hiort et al. J Pediatrics 132: 939-943, 1998
0589 CAIS	Substitut.	6 LBD	*	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T						Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
0599 CAIS	Substitut.	6 LBD	*	775 2323 3438 c.2323C	p.(Arg775 Cys) 3438C >T							Female	Normal		Scheiber et al. J Pedatric Endocrinol & Metab. 16: 367-373,
0731 CAIS	Substitut.	6 LBD	*	775 2324 3439 c.2324G	p.(Arg775 His) 3439G >A						sister affected	Female	Normal	pos	Ledig et al: Horm Res 63: 263-269, 2005
0732 CAIS	Substitut.	6 LBD	*	775 2324 3439 c.2324G	p.(Arg775 His) 3439G >A						mother heterozygous	Female	Normal	pos	Ledig et al: Horm Res 63: 263-269, 2005
0182 CAIS	Substitut.	6 LBD	*	775 2324 3439 c.2324G	p.(Arg775 His) 3439G >A			low	normal	*		Female	Normal		Batch et al. Hum Mol Genet, 1: 497, 1992

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Thermolabile: k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0183	CAIS	Substitut.	**	775	2324	p.(Arg775 His)			v low	high				Female	Normal		DeBellis et al. Mol Endocrinol, 6: 1909-20, 1992
		6 LBD		3439	c.2324G	3439G >A											
0184	CAIS	Substitut.	*	775	2324	p.(Arg775 His)								Female	Normal		Hiort et al. Am J Med Genet. 63. 218-222, 1996
		6 LBD		3439	c.2324G	3439G >A											
0461	CAIS	Substitut.	*	775	2324	p.(Arg775 His)			zero					Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		6 LBD		3439	c.2324G	3439G >A											
0462	CAIS	Substitut.	*	775	2324	p.(Arg775 His)								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		6 LBD		3439	c.2324G	3439G >A											
0181	CAIS	Substitut.	**	775	2324	p.(Arg775 His)			normal	high	high	*	mutation found in two unrelated families	Female	Normal	pos	Prior et al. Am J Hum genet, 51: 143, 1992
		6 LBD		3439	c.2324G	3439G >A											
0863	CAIS	Substitut.	*	775	2323	p.(Arg775 Cys)							Prenat diagnosis bilateral gonadectomy at 5yrs	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
		6 LBD		3438	c.2323C	3438C >T											
0878	CAIS	Substitut.	*	775	2323	p.(Arg775 Cys)								Female	Normal	pos	Qin et al. Zhonghua Fu Chan Ke Za Za 43: 828 -830, 2008
		6 LBD		3438	c.2323C	3438C >T											
0986	CAIS	Substitut.	*	775	2323	p.(Arg775 Cys)	27	17						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		6 LBD		3438	c.2323C	3438C >T											
0987	CAIS	Substitut.	*	775	2324	p.(Arg775 His)	20	17						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		6 LBD		3439	c.2324G	3439G >A											
0185	PAIS	Substitut.	*	775	2324	p.(Arg775 His)											Quigley et al. Endocrin Reviews 16: 271, 1995
		6 LBD		3439	c.2324G	3439G >A											
0186	CAIS	Substitut.	*	780	2338	p.(Arg780 Trp)								Female	Normal		Hiort et al. Hum Mol Genet 3: 1163-1166 1994
		6 LBD		3453	c.2338C	3453C >T											
0187	CAIS	Substitut.	**	780	2338	p.(Arg780 Trp)								Female	Normal		Morono et al. Human Mutation 6: 152-162, 1995
		6 LBD		3453	c.2338C	3453C >T											
0188	CAIS	Substitut.	*	780	2338	p.(Arg780 Trp)								Female	Normal		Sinnecker et al. Eur J. Pediatr. 156: 7-14, 1997
		6 LBD		3453	c.2338C	3453C >T											
0463	CAIS	Substitut.	*	780	2338	p.(Arg780 Trp)								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		6 LBD		3453	c.2338C	3453C >T											
0621	CAIS	Substitut.	*	780	2338	p.(Arg780 Trp)	21		low				AR partially purified appears truncated (43 kD)	Female	Normal		MacLean et al. Hum Mutat. 23: 287, 2004
		6 LBD		3453	c.2338C	3453C >T											
0638	CAIS	Substitut.	*	780	2338	p.(Arg780 Trp)	24						Testis located in inguinal region	Female	Normal	neg	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
		6 LBD		3453	c.2338C	3453C >T											
0733	CAIS	Substitut.	*	780	2338	p.(Arg780 Trp								Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
		6 LBD		3453	c.2338C	3453C >T											
0189	PAIS	Substitut.	*	781	2343	p.(Met781 Ile)			normal	high		*		Female	Ambiguous		Bevan et al. Hum Mol Genet, 5: 265-273, 1996
		6 LBD		3458	c.2343G	3458G >A											
0190	PAIS	Substitut.		781	2343	p.(Met781 Ile)	20	23	normal	high	high		1 family member - male. Rest of family - females	Female / Male	Ambiguous	pos	Pinsky et al. Clin Inv Med, 15: 456, 1992
		6 LBD		3458	c.2343G	3458G >A											
0191	PAIS	Substitut.		781	2343	p.(Met781 Ile)											Brinkmann et al. J Steroid Biochem & Mol Biol 53: 443, 1995
		6 LBD		3458	c.2343G	3458G >A											
0192	PAIS	Substitut.		781	2343	p.(Met781 Ile)							A brother to mutation 0305	Male	Ambiguous	pos	Rodien et al. J Clin Endo & Metab 81: 2904 -2908, 1996
		6 LBD		3458	c.2343G	3458G >A											
0305	CAIS	Substitut.		781	2343	p.(Met781 Ile)							2 sisters to mutation 0192	Female	Normal	pos	Rodien et al. J Clin Endo & Metab 81: 2904 -2908, 1996
		6 LBD		3458	c.2343G	3458G >A											
0193	CAIS	Substitut.		781	2343	p.(Met781 Ile)								Female	Normal		Jakubiczka et al. Human Mutation 9: 57 -61, 1997
		6 LBD		3458	c.2343G	3458G >A											
0464	CAIS	Substitut.		781	2343	p.(Met781 Ile)			low	high			Epididymis & Vas deferens present - Cousin 660	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		6 LBD		3458	c.2343G	3458G >A											
0660	CAIS	Substitut.		781	2343	p.(Met781 Ile)							Epididymis & Vas deferens present -Cousin 464	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		6 LBD		3458	c.2343G	3458G >A											

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0682	PAIS	Substitut.		781	2343	p.(Met781 Ile)								Ambiguous		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
		6 LBD		3458	c.2343G	3458G >A										
0743	PAIS	Substitut.		781	2343	p.(Met781 Ile)						2 affected siblings	Female	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
		6 LBD		3458	c.2343G	3458G >A										
0774	MAIS	Substitut.		781	2343	p.(Met781 Ile)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		6 LBD		3458	c.2343G	3458G >A										
0988	CAIS	Substitut.		782	2344	p.(Tyr782 Asp)	20	18	low	high		Mother & sister heterozygotes, 2 aunts wt	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		6 LBD		3459	c.2344T	3459T >G										
0194	Prostate cancer	Substitut.		783	2348	p.(Ser783 Asn)						Somatic mutation	Male	Normal		Tilley et al. 2: Clinical Cancer Res. 2: 277-285, 1996
		6 LBD		3463	c.2348G	3463G >A										
0383	CAIS	Substitut.	*	785	2354	p.(Cys785 Tyr)			zero			No transactivation capacity	Female	Normal		Giwercman et al. Human Genetics 103: 529-531, 1998
		6 LBD		3469	c.2348G	3469G >A										
0864	CAIS	Substitut.		785	2354	p.(Cys785 Tyr)						Diagnosis & bilateral gonadectomy at 16yrs	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
		6 LBD		3469	c.2348G	3469G >A										
0667	CAIS	Deletion		786	2356	p.(Val786 fs)						Somatic mosaicism 2 nt del causing fs & stop at codon 828	Female	Normal		Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
		6 LBD		3471	c.2356_2357	3471_3472 delGT										
0989	CAIS	Deletion		786	2356	p.(Val786 fs)	19	18	zero	zero		2 nt del causing fs & stop at codon 828	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		6 LBD		3471	c.2356_2357	3471_3472 delGT										
0195	CAIS	Substitut.	*	787	2359	p.(Arg787 *)			zero				Female	Normal		Pinsky et al. Clin Inv Med, 15: 456-472, 1992
		6 LBD		3474	c.2359C	3474C >T										
0557	CAIS	Substitut.	*	787	2359	p.(Arg787 *)							Female	Normal		Ignaccack et al. J Appl Genet 43: 109-114, 2002
		6 LBD		3474	c.2359C	3474C >T										
0600	CAIS	Substitut.	*	787	2359	p.(Arg787 *)							Female	Normal		Scheiber et al. J Pedatric Endocr Metab 16: 367-373, 2003
		6 LBD		3474	c.2359C	3474C >T										
0892	Prostate Cancer	Substitut.	*	787	2359	p.(Arg787 *)						Also p.(Q86del, Q867*. Not treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		6 LBD		3474	c.2359C	3474C >T										
0939	Prostate Cancer	Substitut.	*	787	2359	p.(Arg787 *)						Untreated with anti-androgens	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
		6 LBD		3474	c.2359C	3474C >T										
0196	CAIS	Substitut.	*	788	2362	p.(Met788 Val)			zero				Female	Normal	pos	Nakao et al. J Clin Endocrinol Metab, 74: 1152, 1992
		6 LBD		3477	c.2362A	3477A >G										
0615	CAIS	Substitut.		788	2362	p.(Met788 Val)						Uterus present	Female	Normal		Van et al. Eur J. Pediatr. 162: 781-784, 2003
		6 LBD		3477	c.2362A	3477A >G										
0990	CAIS	Substitut.		788	2364	p.(Met788 Ile)	23	18					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		6 LBD		3479	c.2364G	3479G >A										
0991	CAIS	Substitut.		788	2364	p.(Met788 Ile)	23	17				1 sister affected	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		6 LBD		3479	c.2364G	3479G >A										
0992	CAIS	Substitut.		788	2364	p.(Met788 Ile)	26	17					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		6 LBD		3479	c.2364G	3479G >T										
0406	MAIS	Substitut.		789	2367	p.(Arg789 Ser)	24	23	normal	normal	high	* Gynecomastia -high pitched voice-decreased body hair	Male	Normal	pos	Lumbroso et al 81st. US Endo Soc Meetings Abstr. P3-288, 1999
		6 LBD		3482	c.2367G	3482G >T										
0716	PAIS	Substitut.		789	2367	p.(Arg789 Ser)				high			Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
		6 LBD		3482	c.2367G	3482G >T										
0735	CAIS	Duplicat		789	2365	p.(Arg789 dup)						p.(Arg789) duplicated	Male	Normal	neg	Ledig et al. Horm Res 63: 263-269, 2005
		6 LBD		3480	c.2365_2367	3480_3482 dupAGG										
0197	MAIS	Substitut.	*	791	2371	p.(Leu791 Phe)			normal	low		*	Male	Near-normal male		Tsukada et al: J Clin Endocrinol Metab, 79: 1202, 1994
		6 LBD		3486	c.2371C	3486C >T										
0842	CAIS	Substitut.		791	2372	p.(Leu791 Pro)							Female	Normal		Raicu et al. Asian J Androl 10: 687-91, 2008
		6 LBD		3487	c.2372T	3487T >C										
0993	CAIS	Deletion		793	2378	p.(Gln793 fs)	20	17				Heterozygous sister 2nt del causing fs & stop in Codon 828	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		6 LBD		3493	c.2378_2379	3493_3494 delAA										

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base / HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0198 MAIS	Substitut.	6 LBD		794 2382 3497 c.2382G	p.(Glu794 Asp) 3497G >C			normal	normal		Inconsistent increases in gynocomastia & infertility	Male	Normal		Pinsky et al. Clin Inv Med, 15: 456, 1992
0397 Normal	Substitut.	6 LBD		794 2382 3497 c.2382G	p.(Glu794 Asp) 3497G >C						Homsosexual individual	Male	Normal		Macke et al. Am J Human Genetics 53: 844-852, 1993
0199 CAIS	Substitut.	6 LBD		795 2384 3499 c.2384T	p.(Phe795 Ser) 3499T >C			zero				Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0200 CAIS	Substitut.	6 LBD		795 2384 3499 c.2384T	p.(Phe795 Ser) 3499T >C							Female	Normal		Jakubiczka et al Human Mutation 9: 57-61, 1997
0907 PAIS	Substitut.	6 LBD		796 2388 3503 c.2388A	p.(Gly796 Gly) 3503A >G			low	high		Silent mutation	Male	Ambiguous		Appari et al. J Mol Med 87: 623-632, 2009
0201 CAIS	Substitut.	6 LBD	*	797 2391 3506 c.2391G	p.(Trp797 *) 3506G >A			v low				Female	Normal		Marcelli at al. J Clin Invest 85: 1522-1528, 1990
0202 PAIS	Substitut.	6 LBD	*	799 2395 3510 c.2395C	p.(Gln799 Glu) 3510C >G			normal	normal	*		Female	Ambiguous		Bevan et al. Hum Mol Genet, 5: 265-273, 1996
0203 PAIS	Substitut.	6 LBD		799 2395 3510 c.2395C	p.(Gln799 Glu) 3510C >G			normal	normal						Quigley et al. Endocrine Reviews 16: 271, 1995
0204 PAIS	Substitut.	6 LBD		799 2395 3510 c.2395C	p.(Gln799 Glu) 3510C >G							Female	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0205 Prostate cancer	Substitut.	6 LBD		799 2395 3510 c.2395C	p.(Gln799 Glu) 3510C >G						Also present in genomic DNA	Male	Normal		Evans et al. Prostate 28: 162-171, 1996
0399 Prostate cancer	Substitut.	6 LBD		799 2395 3510 c.2395C	p.(Gln799 Glu) 3510C >G						Somatic mutation Stage 4 tumor	Male	Normal		Castagnaro et al. Verh. Dtsch. Ges. Path. 77. 119-123, 1993
0340 MAIS	Substitut.	6 LBD	*	799 2395 3510 c.2395C	p.(Gln799 Glu) 3510C >G			normal			Azospermia	Male	Normal		Hiort et al. J Clin Endocrinol Metab 85: 2810-2815, 2000
0381 MAIS	Substitut.	6 LBD	*	799 2395 3510 c.2395C	p.(Gln799 Glu) 3510C >G			normal			Azoospermia - defective transactivation	Male	Normal		Wang et al. J Clin Endocrinol Metab 83: 4303-4309, 1998
0775 MAIS	Substitut.	6 LBD		799 2395 3510 c.2395C	p.(Gln799 Glu) 3510C >G						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
0954 PAIS	Substitut.	6 LBD	*	799 2395 3510 c.2395C	p.(Gln799 Glu) 3510C >G						Virilization post-gonadectomy heteroz P450 (POR) p.Y601C	Female	Ambiguous		Idkowiak et al. J Clin Endocrinol Metab 95: 3418-3527, 2010
1041 PAIS	Substitut.	6 LBD	*	799 2395 3510 c.2395C	p.(Gln799 Glu) 3510C >G						External Masculisation Score = 8	Male	Ambiguous		Hellmann et al. Arch Dis Child 97:403-409, 2012
0648 CAIS	Substitut.	6 LBD		799 2395 3510 c.2395C	p.(Gln799 *) 3510C >T						Abdominal tumor - Sertoli cell adenoma	Female	Normal		Ignacek et al: Gynecol Endorinol 19: 178-181, 2004
0764 Prostate cancer	Substitut.	6 LBD		800 2399 3514 c.2399T	p.(Ile800 Thr) 3514T >C						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006
0596 PAIS	Substitut.	6 LBD		801 2402 3517 c.2402C	p.(Thr801 Ile) 3517C >T						Variable expressity-four additional males with MAIS	Male	Ambiguous	pos	Galli-Tsinopoulou et al. J Pediatr Endocrinol Metab16: 149-154,
0542 CAIS	Deletion	6 LBD		801 2403 3518 c.2403	p.(Thr801 fs) 3518 delC						Single nt del causing frameshift & stop in codon 808 1 affect sib	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
1055 CAIS	Deletion	6 LBD		801 2403 3518 c.2403	p.(Thr801 fs) 3518 delC						Single nt del causing frameshift & stop in codon 808 1 affect sib	Female	Normal	pos	Vasu et al. Genet Test Mol Biomarkers 16:749 -755, 2012
0521 PAIS	Substitut.	6 LBD		803 2408 3523 c.2408A	p.(Gln803 Arg) 3523A >G										Chavez et al. Clin Genet 59: : 185-188, 2001
0498 CAIS	Substitut.	6 LBD		804 2410 3525 c.2410G	p.(Glu804 Lys) 3525G >A			zero	zero			Female	Normal	pos	Sawai et al. J Hum Genet 45: 342-345, 2000
0563 CAIS	Substitut.	6 LBD		805 2413 3528 c.2413T	p.(Phe805 Leu) 3528T >C						*de novo* mutation. + glaucoma & hyper-trophic pyloric stenos	Female	Normal	neg	Gad et al. Clin Genet. 63: 59-63, 2003
0865 CAIS	Substitut.	6 LBD		805 2413 3528 c.2413T	p.(Phe805 Ile) 3528T >A						Diagnosis at 1mo	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008

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0206	PAIS	Substitut.		6 LBD	807 3535	2420 c.2420G	p.(Cys807 Tyr) 3535G >A									Brown et al. Eur J Pediatr 152: (Suppl 2) S62, 1993
0207	CAIS	Substitut.	*	6 LBD	808 3537	2422 c.2422A	p.(Met808 Val) 3537A >G			low			Female	Normal		Morono et al. Human Mutation 6: 152-162, 1995
0639	CAIS	Substitut.		6 LBD	808 3537	2422 c.2422A	p.(Met808 Val) 3537A >G	30		low		Testis located in inguinal region	Female	Normal	neg	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
0428	PAIS	Substitut.	*	6 LBD	808 3538	2423 c.2423T	p.(Met808 Thr) 3538T >C			low		Treatment with topical DHT restored male genital	Female	Ambiguous		Ong et al. Lancet 354: 1444-1445, 1999
0208	CAIS	Substitut.		6 LBD	808 3538	2423 c.2423T	p.(Met808 Arg) 3538T >G			zero			Female	Normal		Adeyemo et al. Hum Mol Genet, 2: 1809, 1993
1093	CAIS	Substitut.		6 LBD	809 3540	2425 c.2425A	p.Lys809 Gln) 3540A >C						Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0403	PAIS	Substitut.		6 LBD	813 3552	2437 c.2437C	p.(Leu813 Phe) 3552C >T						Female	Normal		Yaegashi et al. Tohoku J of Exp Med 187: 263 -272, 1999
0799	CAIS	Substitut.	*	6 LBD	813 3553	2438 c.2438T	p.(Leu813 Pro) 3553T >C			zero		Severely impaired AR transactivation	Female	Normal		Jaaskelainen et al. Hum Mutat 27: 291, 2006
0209	PAIS	Substitut.		6 LBD	815 3559	2444 c.2444G	p.(Ser815 Asn) 3559G >A	20		normal		Hormone binding specificity alterted.	Female	Ambiguous		Pinsky et al. Clin Inv Med, 15: 456, 1992
0210	MAIS	Substitut.		6 LBD	815 3559	2444 c.2444G	p.(Ser815 Asn) 3559G >A	20		normal		Hormone binding specificity altered	Male	Normal	pos	Pinsky et al. Clin Inv Med, 15: 456, 1992
0685	PAIS	Substitut.		7 LBD	818 3567	2452 c.2452C	p.(Pro818 Ala) 3567C >G			normal high		Mother a carrier	Male	Ambiguous	pos	Lumbroso et al. J Mol Endocrinol 32: 679 -687, 2004
0811	CAIS	Substitut.		7 LBD	818 3568	2453 c.2453C	p.(Pro818 Leu) 3568C >T					Two sisters with CAIS mother a carrier	Female	Normal	pos	Turek-Plewa et al. Hum Genet 119: 361, 2006
0501	CAIS	Substitut.		7 LBD	820 3574	2459 c.2459A	p.(Asp820 Gln) 3574A >G					Also 3578Gdel causing frameshift & stop in codon 821	Female	Normal		Choi et al. Arch Gynecol Obstet 263: 201-205, 2000
0211	CAIS	Substitut.	*	7 LBD	821 3577	2462 c.2462G	p.(Gly821 Ala) 3577G >C			normal high	*	Also Leu263Pro, enhances thermolability	Female	Ambiguous	neg	Tanaka et al. Gynecological Endo. 12: 75-82, 1998
0212	PAIS	Substitut.		7 LBD	822 3579	2464 c.2464C	p.(Leu822 Val) 3579C >G	24	23	normal normal						Pinsky et al. Clin Inv Med, 15: 456, 1992
0776	MAIS	Substitut.		7 LBD	822 3579	2464 c.2464C	p.(Leu822 Val) 3579C >G					Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
0513	MAIS	Substitut.	*	7 LBD	825 3588	2473 c.2473C	p.(Gln825 Lys) 3588C >A					Gynocomastia-norm fert -related to 514 abnormal Bmax DHT	Male	Normal	pos	Giwercman et al. J Clin Endocrinol Metab 85: 2253-2259, 2000
0514	MAIS	Substitut.	*	7 LBD	825 3588	2473 c.2473C	p.(Gln825 Lys) 3588C >A					Gynocomastia-norm fert -related to 513 abnormal Bmax DHT	Male	Normal	pos	Giwercman et al. J Clin Endocrinol Metab 85: 2253-2259, 2000
1041	PAIS	Substitut.	*	7 LBD	825 3588	2473 c.2473C	p.(Gln825 Lys) 3588C >A					External Masculisation Score = 12	Male	Ambiguous		Hellmann et al. Arch Dis Child 97:403-409, 2012
1042	PAIS	Substitut.	*	7 LBD	825 3588	2473 c.2473C	p.(Gln825 Lys) 3588C >A					External Masculisation Score = 12	Male	Ambiguous	pos	Hellmann et al. Arch Dis Child 97:403-409, 2012
1043	PAIS	Substitut.	*	7 LBD	825 3588	2473 c.2473C	p.(Gln825 Lys) 3588C >A					External Masculisation Score = 12	Male	Ambiguous		Hellmann et al. Arch Dis Child 97:403-409, 2012
1044	PAIS	Substitut.	*	7 LBD	825 3588	2473 c.2473C	p.(Gln825 Lys) 3588C >A					External Masculisation Score = 12	Male	Ambiguous		Hellmann et al. Arch Dis Child 97:403-409, 2012
1045	PAIS	Substitut.	*	7 LBD	825 3588	2473 c.2473C	p.(Gln825 Lys) 3588C >A					External Masculisation Score =12-brother has PAIS	Male	Ambiguous	pos	Hellmann et al. Arch Dis Child 97:403-409, 2012
1046	PAIS	Substitut.	*	7 LBD	825 3588	2473 c.2473C	p.(Gln825 Lys) 3588C >A					External Masculisation Score = 10, cousins PAIS	Male	Ambiguous	pos	Hellmann et al. Arch Dis Child 97:403-409, 2012
1047	PAIS	Substitut.	*	7 LBD	825 3588	2473 c.2473C	p.(Gln825 Lys) 3588C >A					External Masculisation Score = 12, cousins PAIS	Male	Ambiguous	pos	Hellmann et al. Arch Dis Child 97:403-409, 2012

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0875 PAIS	Substitut.	7 LBD	*	827 3596	2481 c.2481C	p.(Phe827 Leu) 3596C >A	22	17	normal			Increased N/C terminal interaction & TIF2 co-activation	Male	Ambiguous	pos	Wong et al. Mol Cell Endocrinol 292: 69-78, 2008
0537 CAIS	Substitut.	7 LBD		828 3597	2482 c.2482T	p.(Phe828 Val) 3597T >G							Female	Normal		Chavez et al. J Hum Genet. 46: 560-565, 2001
1094 CAIS	Substitut.	7 LBD		828 3598	2483 c.2483T	p.(Phe828 Ser) 3598T >C							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0622 CAIS	Insertion	7 LBD		830 3593	2478 c.2478_2479	p.(Glu830 fs) 3603_3604 insC A	18					2nt insertion causing frameshift and stop in Codon 834	Female	Normal	pos	MacLean et al. Hum Mutat. 23: 287, 2004
0522 CAIS	Substitut.	7 LBD		831 3606	2491 c.2494C	p.(Leu831 Val) 3606C >G							Female	Normal		Chavez et al. Clin Genet 59: : 185-188, 2001
0213 CAIS	Substitut.	7 LBD	*	832 3609	2494 c.2494C	p.(Arg832 *) 3609C >T			zero				Female	Normal	pos	DeBellis et al. Mol Endocrinol, 6: 1909-20, 1992
0214 CAIS	Substitut.	7 LBD	*	832 3609	2494 c.2494C	p.(Arg832 *) 3609C >T			zero				Female	Normal		Tincello et al. J Endocrinol, 132 Suppl, Abstr 87, 1992
0215 CAIS	Substitut.	7 LBD	*	832 3609	2494 c.2494C	p.(Arg832 *) 3609C >T			zero				Female	Normal		Ris-Stalpers et al. 74th Endo Soc Meeting, 1992
0384 CAIS	Substitut.	7 LBD	*	832 3609	2494 c.2494C	p.(Arg832 *) 3609C >T							Female	Normal		Giwercman et al. Human Genetics 103: 529-531, 1998
0465 CAIS	Substitut.	7 LBD	*	832 3609	2494 c.2494C	p.(Arg832 *) 3609C >T						Wolffian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0500 CAIS	Substitut.	7 LBD	*	832 3609	2494 c.2494C	p.(Arg832 *) 3609C >T							Female	Normal		Choi et al. Arch Gynecol Obstet 263: 201-205, 2000
0515 CAIS	Substitut.	7 LBD	*	832 3609	2494 c.2494C	p.(Arg832 *) 3609C >T						Harmatoma found in pubertal patient	Female	Normal		Chen et al. Fertilty & Sterility 74: 182-183, 2000
0466 CAIS	Substitut.	7 LBD	*	832 3610	2495 c.2495G	p.(Arg832 Gln) 3610G >A							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0499 CAIS	Substitut.	7 LBD	*	832 3610	2495 c.2495G	p.(Arg832 Gln) 3610G >A							Female	Normal		Choi et al. Arch Gynecol Obstet 263: 201-205, 2000
0814 CAIS	Substitut.	7 LBD	*	832 3610	2495 c.2495G	p.(Arg832 Gln) 3610G >A						"hamatomatous testes" present	Female	Normal		Goulis et al. Hirmines (Athens) 5: 200-204, 2006
0216 CAIS	Substitut.	7 LBD	**	832 3610	2495 c.2495G	p.(Arg832 Gln) 3610G >A			v low				Female	Normal	pos	Brown et al. Mol Endocrinol, 4: 1759-72, 1990
0217 CAIS	Substitut.	7 LBD	*	832 3610	2495 c.2495G	p.(Arg832 Gln) 3610G >A			zero			Found in two unrelated families	Female	Normal		McPhaul et al. J Clin Inv, 90: 2097, 1992
0404 CAIS	Substitut.	7 LBD		832 3610	2495 c.2495G	p.(Arg832 Gln) 3610G >A			zero				Female	Normal		Yaegashi et al. Tohoku J of Exp Med 187: 263 -272, 1999
0524 CAIS	Substitut.	7 LBD		832 3610	2495 c.2495G	p.(Arg832 Gln) 3610G >A			zero			Sertoli cell carcinoma	Female	Normal		Ko et al. Int. J. Gynocol. Pathol. 20: 196-199, 2001
0994 CAIS	Substitut.	7 LBD		832 3610	2495 c.2495G	p.(Arg832 Gln) 3610G >A	23	17				Mother heterozygous carrier	Female	Normal		Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0218 CAIS	Substitut.	7 LBD	*	832 3610	2495 c.2495G	p.(Arg832 Leu) 3610G >T	21	19	zero				Female	Normal		Shkolny et al. Human Mol Genetics 4: 515 -521,1995
0307 CAIS	Substitut.	7 LBD	*	832 3610	2495 c.2495G	p.(Arg832 Leu) 3610G >T	26	16	zero				Female	Normal		Shkolny et al. Human Mol Genetics 4: 515 -521,1995
0590 CAIS	Substitut.	7 LBD	*	832 3610	2495 c.2495G	p.(Arg832 Leu) 3610G >T						Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
0995 CAIS	Substitut.	7 LBD	*	832 3609	2494 c.2494C	p.(Arg832 *) 3609C >T	19	18					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1095 CAIS	Substitut.	7 LBD				p.(Met833 *)							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012

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0996	CAIS	Deletion	7 LBD	834 / 3615	2500 / c.2500_2502	p.(Asn834 del) / 3615_3617 delAAC	19	18					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0219	CAIS	Substitut.	7 LBD	835 / 3619	2504 / c.2504A	p.(Tyr835 Cys) / 3619A >G			zero				Female	Normal		Wilson et al. J Clin Endocrinol Metab, 75: 1474-8, 1992
0392	PAIS	Substitut.	7 LBD	839 / 3632	2517 / c.2517C	p.(Leu839) / 3632C >T						Hypospadia and cryptorchidism - silent mutation	Male	Ambiguous		Nordenskjold et al Urological Res, 27: 49 -55, 1999
0765	Prostate cancer	Substitut.	7 LBD	839 / 3632	2517 / c.2517C	p.(Leu839) / 3632C >T						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006
0997	CAIS	Substitut.	7 LBD	839 / 3630	2515 / c.2515C	p.(Leu839 Val) / 3630C >G	8	17					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1038	PAIS	Substitut.	7 LBD	839 / 3630	2515 / c.2515C	p.(Leu839 Phe / 3630C >T						External Masculisation Score = 6	Male	Ambiguous		Hellmann et al. Arch Dis Child 97:403-409, 2012
0220	PAIS	Substitut.	7 LBD	** 841 / 3636	2521 / c.2521C	p.(Arg841 Cys) / 3636C >T	20	16	normal	high	norm *		Male	Ambiguous	pos	Beitel et al. J Clin Inv, 94: 546-554 1994
0221	PAIS	Substitut.	7 LBD	** 841 / 3636	2521 / c.2521C	p.(Arg841 Cys) / 3636C >T			low	high	*	Found in two unrelated individuals.	Female			McPhaul et al. J Clin Inv, 90: 2097, 1992
0222	PAIS	Substitut.	7 LBD	** 841 / 3636	2521 / c.2521C	p.(Arg841 Cys) / 3636C >T			normal	high		Sibling of 0308	Female	Ambiguous	pos	Bevan et al. Hum Mol Genet, 5: 265-273, 1996
0308	PAIS	Substitut.	7 LBD	** 841 / 3636	2521 / c.2521C	p.(Arg841 Cys) / 3636C >T			normal	high		Sibling of 0222	Male	Ambiguous	pos	Bevan et al. Hum Mol Genet, 5: 265-273, 1996
0387	PAIS	Substitut.	7 LBD	** 841 / 3636	2521 / c.2521C	p.(Arg841 Cys) / 3636C >T						Transcriptional activity only at high conc of androgen				Georget et al. J Clin Endocrinol Metab 83: 3597-3603, 1998
0698	PAIS	Substitut.	7 LBD	** 841 / 3636	2521 / c.2521C	p.(Arg841 Cys) / 3636C >T			normal	low		Family member of 0699 & 0700 with phenotypic varaiation	Male	Ambiguous	pos	Wang et al. Biochem Biophs Res Comm 335: 335-342, 2005
0699	PAIS	Substitut.	7 LBD	** 841 / 3636	2521 / c.2521C	p.(Arg841 Cys) / 3636C >T			normal	low		Family member of 0698 & 0700 with phenotypic varaiation	Male	Ambiguous	pos	Wang et al. Biochem Biophs Res Comm 335: 335-342, 2005
0700	PAIS	Substitut.	7 LBD	** 841 / 3636	2521 / c.2521C	p.(Arg841 Cys) / 3636C >T			normal	low		Family member of 0698 & 0699 with , phenotypic varaiation	Male	Ambiguous	pos	Wang et al. Biochem Biophs Res Comm 335: 335-342, 2005
0734	PAIS	Substitut.	7 LBD	* 841 / 3636	2521 / c.2521C	p.(Arg841 Cys) / 3636C >T						brother affected, sister heterozygous	Male	Ambiguous	pos	Ledig et al: Horm Res 63: 263-269, 2005
0385	PAIS	Substitut.	7 LBD	* 841 / 3636	2521 / c.2521C	p.(Arg841 Gly) / 3636C >G			low			Reduced transactivation				Giwercman et al. Human Genetics 103: 529-531, 1998
0818	PAIS	Substitut.	7 LBD	* 841 / 3637	2522 / c.2522G	p.(Arg841 His) / 3637G >A							Male	Ambiguous		Yen et al. Acta Paediatr Taiwan 46: 101-105, 2005
0683	CAIS	Substitut.	7 LBD	* 841 / 3637	2522 / c.2522G	p.(Arg841 His) / 3637G >A						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0820	CAIS	Substitut.	7 LBD	* 841 / 3637	2522 / c.2522G	p.(Arg841 His) / 3637G >A							Female	Normal		Wang et al. Yi Chuan Xue Bao 33: 19-25, 2006
0337	PAIS	Substitut.	7 LBD	** 841 / 3637	2522 / c.2522G	p.(Arg841 His) / 3637G >A	19		normal	high	high *		Female	Ambiguous	pos	Beitel et al. J Clin Inv, 94: 546-554 1994
0224	PAIS	Substitut.	7 LBD	** 841 / 3637	2522 / c.2522G	p.(Arg841 His) / 3637G >A	18	24	normal	high	high *		Female	Ambiguous	pos	Beitel et al. J Clin Inv, 94: 546-554 1994
0225	PAIS	Substitut.	7 LBD	* 841 / 3637	2522 / c.2522G	p.(Arg841 His) / 3637G >A				high		Found in two unrelated families	Female	Ambiguous	pos in 1 fam	Hiort et al. J Clin Endocrinol Metab, 77: 262-266, 1993
0226	PAIS	Substitut.	7 LBD	* 841 / 3637	2522 / c.2522G	p.(Arg841 His) / 3637G >A			zero							McPhaul et al. J Clin Inv, 90: 2097, 1992
0227	PAIS	Substitut.	7 LBD	* 841 / 3637	2522 / c.2522G	p.(Arg841 His) / 3637G >A			normal	normal	*	In same fam. persons raised as males with ambiguous genitalia	Female	Ambiguous	pos	Imasaki et al. Eur J Endorinol, 130: 569 -574, 1994
0228	PAIS	Substitut.	7 LBD	* 841 / 3637	2522 / c.2522G	p.(Arg841 His) / 3637G >A			low							Lumbroso et al. Eur J Endorinol 130: 327, 1994

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0229 PAIS	Substitut.	7 LBD	*	841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A			low							Imai et al. Annals of Clinical Biochem, 32: 482-486, 1995
0230 PAIS	Substitut.	7 LBD	*	841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A										Ghirri & Brown. Pediatr Res 33: Abstr.95, 1993
0231 PAIS	Substitut.	7 LBD	**	841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A			low	high	high					Marcelli et al. J Clin Invest 94: 1642-1650, 1994
0232 PAIS	Substitut.	7 LBD	**	841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A			normal	high		*	Female	Ambiguous	pos	Weidemann et al. Clin Endocrinology 45: 733 -
0223 PAIS	Substitut.	7 LBD	**	841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A			low	high			Female	Ambiguous		De Bellis et al. J Clin Endrcrinol Metab, 78: 513, 1994
0668 PAIS	Substitut.	7 LBD	*	841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A						Somatic mosaicism	Male	Ambiguous		Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
0415 PAIS	Substitut.	7 LBD		841 3636	2521 c.2521C	p.(Arg841 Ser) 3636C >A	25					Testis located in inguinal region. same family as 0832	Male	Ambiguous		Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
0626 PAIS	Substitut.	7 LBD		841 3636	2521 c.2521C	p.(Arg841 Ser) 3636C >A						Diagnosis at 6.5 yr. Reassigned as male after testosterone	Female	Ambiguous		Mazen et al. J Endocrinol Invest. 27: 57-60, 2004
0832 PAIS	Substitut.	7 LBD		841 3636	2521 c.2521C	p.(Arg841 Ser) 3636C >A						Testis located in inguinal region. same family as 0415	Male	Ambiguous		Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
1008 PAIS	Substitut.	7 LBD	*	841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A	23	11				1 affected niece	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0233 PAIS	Substitut.	7 LBD		842 3640	2525 c.2525T	p.(Ile842 Ser) 3640T >G			low	high			Female	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0234 CAIS	Substitut.	7 LBD		843 3643	2528 c.2528T	p.(Ile843 Thr) 3643T >C			normal	high			Female	Normal	pos	Hiort et al. J Clin Endocrinol Metab, 77: 262-266, 1993
0235 PAIS	Substitut.	7 LBD	*	843 3643	2528 c.2528T	p.(Ile843 Thr) 3643T >C			low	high		*	Male	Ambiguous	pos	Weidemann et al Clin Endocrinlogy 45: 733 - 739, 1996
0591 CAIS	Substitut.	7 LBD		843 3643	2528 c.2528T	p.(Ile843 Asn) 3643T >A						Normal immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
0736 PAIS	Substitut.	7 LBD		844 3646	2531 c.2531C	p.(Ala844 Glu) 3646C >A						de novo mutation	Male	Ambiguous	neg	Ledig et al. Horm Res 63: 263-269, 2005
0494 Prostate cancer	Substitut.	7 LBD		847 3654	2539 c.2539A	p.(Arg847 Gly) 3654A >G						Somatic mutation	Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000
0236 CAIS	Insertion	7 LBD		849 3661	2546 c.2546_2547	p.(Asn849 fs) 3661_3662 insA			zero			1 nt insert frameshift & stop in codon 880 & loss of 44 AA's	Female	Normal		Brinkmann et al. J Steroid Biochem Mol Biol 53: 443, 1995
0467 CAIS	Insertion	7 LBD		849 3661	2546 c.2546_2547	p.(Asn849 fs) 3661_3662 insA			zero			1 nt insert causes frameshift.	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0854 CAIS	Insertion	7 LBD		849		p.(Asn849 fs)						Frameshift & stop in Codon 880 Diag & bilat gonadect at 7y	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
0957 CAIS	Insertion	7 LBD		849 3661	2546 c.2546_2547	p.(Asn849 fs) 3661_3662 insA						1 nt insert - frameshift & stop in Codon 880	Female	Normal	neg	Wu et al. Fertility & Sterility 93: 2076, e1-4, 2010
1096 CAIS	Substitut.	7 LBD		849 3663	2548 c.2548T	p.(Asn Lys) 3663T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0237 CAIS	Substitut.	7 LBD		854 3676	2561 c.2561C	p.(Ser854 *) 3676C >G			zero				Female	Normal		Wilson et al. J Clin Endocrinol Metab, 75: 1474-8, 1992
0238 CAIS	Substitut.	7 LBD		854 3676	2561 c.2561C	p.(Ser854 *) 3676C >G			zero				Female	Normal		Jakubiczka et al. Human Mutation 9: 57 -61, 1997
0793 CAIS	Deletion	7 LBD		854 3677	2562 c.2562_2563	p.(Ser854) 3677_3678 delAA						2 nt deletion	Female	Normal		Mueller et al. Hum Genet 119: 673, 2006
0239 PAIS	Substitut.	7 LBD		855 3679	2564 c.2564G	p.(Arg855 Lys) 3679G >A			low			*				McPhaul et al. J Clin Inv, 90: 2097, 1992

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1056	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)							Female	Normal		Vasu et al. Genet Test Mol Biomarkers 16:749 -755, 2012
		7 LBD		3681	c.2566C	3681C >T										
0240	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)			zero				Female	Normal		DeBellis et al. Mol Endocrinol 6: 1909-20, 1992
		7 LBD		3681	c.2566C	3681C >T										
0241	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)							Female	Normal		Tincello et al. J Endocrinol 132 Suppl, Abstr 87, 1992
		7 LBD		3681	c.2566C	3681C >T										
0242	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)			zero				Female	Normal		McPhaul et al. J Clin Inv, 90: 2097, 1992
		7 LBD		3681	c.2566C	3681C >T										
0243	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)							Female	Normal		Loboccaro et al. Pediat Res 33: Abstr 115, 1993
		7 LBD		3681	c.2566C	3681C >T										
0244	CAIS	Substitut.	**	856	2566	p.(Arg856 Cys)			low				Female	Normal	pos	Morono et al. Human Mutation 6: 152-162. 1995
		7 LBD		3681	c.2566C	3681C >T										
0245	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)			zero				Female	Normal		Sultan et al. J Steroid Biochem & Mol Biol: 40
		7 LBD		3681	c.2566C	3681C >T										
0246	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)							Female	Normal		Brinkmann et al. J Steroid Biochem & Mol Biol 53: 443, 1995
		7 LBD		3681	c.2566C	3681C >T										
0247	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)			zero				Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996
		7 LBD		3681	c.2566C	3681C >T										
0248	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)			v low	high			Female	Normal	pos	Malmgren et al.Clin Genet. 50: 202-205, 1996
		7 LBD		3681	c.2566C	3681C >T										
0320	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)							Female	Normal		Komori et al: J Obstetrics & Gynocol. Res. 23: 277-81, 1997
		7 LBD		3681	c.2566C	3681C >T										
0468	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)			zero				Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		7 LBD		3681	c.2566C	3681C >T										
0469	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)			normal	high			Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		7 LBD		3681	c.2566C	3681C >T										
0527	CAIS	Substitut.	**	856	2566	p.(Arg856 Cys)			v low	high			Female	Normal		Elhaji et al. 83rd US Endo Soc Meeting, Abstr P2-37, 2001
		7 LBD		3681	c.2566C	3681C >T										
0592	CAIS	Substitut.	**	856	2566	p.(Arg856 Cys)						v low immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188, 2002
		7 LBD		3681	c.2566C	3681C >T										
0640	CAIS	Substitut.	**	856	2566	p.(Arg856 Cys)	19		zero			Testis located in abdomen Same family as 0833	Female	Normal	pos	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
		7 LBD		3681	c.2566C	3681C >T										
0737	CAIS	Substitut.	*	856	2566	p.(Arg856 Cys)						de novo mutation	Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
		7 LBD		3681	c.2566C	3681C >T										
0661	CAIS	Substitut.	*	856	2567	p.(Arg856 His)						Vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		7 LBD		3682	c.2567G	3682G >A										
0528	PAIS	Substitut.	**	856	2567	p.(Arg856 His)			normal	high	*		Male	Ambiguous		Elhaji et al. 83rd US Endo Soc Meeting, Abstr P2-37, 2001
		7 LBD		3682	c.2567G	3682G >A										
0684	CAIS	Substitut.	*	856	2567	p.(Arg856 His)						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
		7 LBD		3682	c.2567G	3682G >A										
0688	CAIS	Substitut.	*	856	2567	p.(Arg856 His)						Mother a heterozyote	Female	Normal		Skordis et al. J Pediatr Endocrinol Metab 18: 309-313, 2005
		7 LBD		3682	c.2567G	3682G >A										
0745	PAIS	Substitut.	*	856	2567	p.(Arg856 His)						sibling of 0746	Male	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
		7 LBD		3682	c.2567G	3682G >A										
0746	PAIS	Substitut.	*	856	2567	p.(Arg856 His)			normal	high		niece of 0745 also P392S	Female	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
		7 LBD		3682	c.2567G	3682G >A										
0777	MAIS	Substitut.	*	856	2567	p.(Arg856 His)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		7 LBD		3682	c.2567G	3682G >A										
0641	PAIS	Substitut.		856	2567	p.(Arg856 His)	19		zero to normal			Testis located in inguinal region	Male	Ambiguous	neg	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
		7 LBD		3682	c.2567G	3682G >A										

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0716 PAIS	Substitut.	7 LBD		856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A				high			Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
0251 PAIS	Substitut.	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A			normal	high						Chang et al. 73rd Endo Soc Meeting, Abstr 28, 1991
0252 PAIS	Substitut.	7 LBD	**	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A			normal	high		* Servere hypospadia	Male	Ambiguous	pos	Batch et al. Hum Mol Genet, 1: 497, 1992
0253 PAIS	Substitut.	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A							Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222. 1996
0254 PAIS	Substitut.	7 LBD	**	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A			zero				Female	Ambiguous	pos	Weidemann et al. Clin Endocrinology 45: 733 -
0255 PAIS	Substitut.	7 LBD	**	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A			low	high	norm		Female	Ambiguous		Marcelli et al. J Clin Invest, 94: 1642-1650, 1994
0301 PAIS	Substitut.	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	14					Brother of 0302 somatic & germ-line muts. in mother	Male	Ambiguous	pos	Boehmer et al. Am J Hum Genetics 60: 1003 -6, 1997
0250 PAIS	Substitut.	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A			zero				Female	Ambiguous		Weidemann et al. Clin Endorinology 45: 733 -739, 1996
0302 PAIS	Substitut.	7 LBD	**	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	14					Sister of 0301. somatic & germ-line muts. in mother	Female	Ambiguous	pos	Boehmer et al. Am J Hum Genetics 60: 1003 -6, 1997
0249 CAIS	Substitut.	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A			low				Female	Normal		McPhaul et al. J Clin Invest. 90: 2097, 1992
0344 PAIS	Substitut.	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	28		zero to normal			Sex changed at 1yr Male to female - Same family as 0834	Female	Ambiguous	pos	Melo et al. J Clin Endorinol & Metab 88: 3241-3250, 2003
0833 CAIS	Substitut.	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T						gonads located in abdomen. Same family as 0604	Female	Normal		Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
0834 PAIS	Substitut.	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A						Same family as 0344	Male	Ambiguous	pos	Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
0866 CAIS	Substitut.	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T						Diagnosis at 3 wks	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
0998 CAIS	Substitut.	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	20	14				1 affected aunt; mother & 2 aunts heterozygotes	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1009 PAIS	Substitut.	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	18	17				1 affected sister, mot & 2 aunts heterozygote carriers	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0470 CAIS	Substitut.	7 LBD		857 3686	2571 c.2571C	p.(Phe857 Leu) 3686C >G							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0662 CAIS	Substitut.	7 LBD		857 3686	2571 c.2571C	p.(Phe857 Leu) 3686C >G						Also S866P-twin of 0663	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0663 CAIS	Substitut.	7 LBD		857 3686	2571 c.2571C	p.(Phe857 Leu) 3686C >G						Also S866P-twin of 0662	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0356 CAIS	Substitut.	7 LBD		858		p.(Tyr858 *)						de novo mutation	Female	Normal	neg	Hiort et al. J Pediatrics 132: 939-943, 1998
0941 CAIS	Duplicat.	7 LBD		858 3688	2573 c.2573	p.(Tyr858 *) 3688 dupA						1nt duplicatn resulted in stop codon. Mother & sister carriers	Female	Normal	pos	Turek-Plewa et al. Hum Genet 125: 341, 2009
0753 CAIS	Substitut.	7 LBD	*	860 3693	2578 c.2578C	p.(Leu860 Phe) 3693C >T			v low			In silico analysis showed effect on ligand-binding pocket	Female	Normal		Rajender et al. J Andrology 28: 772-6, 2007
0867 CAIS	Substitut.	7 LBD		861 3697	2582 c.2582C	p.(Thr861 Asn) 3697C >A						Diagnosis at birth bilateral gonadectomy at 15yrs	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
1097 CAIS	Substitut.	7 LBD		861 3697	2582 c.2582C	p.(Thr861 Ile) 3697C >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0256 CAIS	Substitut.	7 LBD		864 3706	2591 c.2591T	p.(Leu864 Arg 3706T >G							Female	Normal		Brown et al. Eur J Pediatr 152: (Suppl 2) S62, 1993

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0257	CAIS	Substitut.	*	865	2593	p.(Asp865 Asn)			low			Transactivation activity increases with horm.	Female	Normal		Bevan et al. J Steroid Biochem Mole Biol 61: 19-26, 1997
		7 LBD		3708	c.2593G	3708G >A										
0471	CAIS	Substitut.		865	2593	p.(Asp865 Asn)						Epididymis & vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		7 LBD		3708	c.2593G	3708G >A										
0472	CAIS	Substitut.		865	2594	p.(Asp865 Gly)			zero			Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		7 LBD		3709	c.2594A	3709A >G										
0258	CAIS	Substitut.	*	865	2594	p.(Asp865 Gly)			zero				Female	Normal		DeBellis et al. Mol Endocrinol, 6: 1909-20, 1992
		7 LBD		3709	c.2594A	3709A >G										
0486	CAIS	Substitut.		866	2596	p.(Ser866 Pro)							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		7 LBD		3711	c.2596T	3711T >C										
0560	CAIS	Substitut.		866	2596	p.(Ser866 Pro)						de novo mut. also Phe857Leu mut -no effect horm binding	Female	Normal	pos	Mongan et al. J Clin Endocrinol Metab 87: 1057-1061, 2002
		7 LBD		3711	c.2596T	3711T >C										
0345	PAIS	Substitut.		867	2599	p.(Val867 Leu)	25		normal	high			Male	Ambiguous		Saunders et al. Clin Endocrinol, 37: 214, 1992
		7 LBD		3714	c.2599G	3714G >T										
0260	PAIS	Substitut.	*	867	2599	p.(Val867 Leu)			normal	high			Male	Ambiguous	pos	Kazemi-Esfarjani et al. Mol Endocrinol, 7: 37 -46, 1993
		7 LBD		3714	c.2599G	3714G >T										
0261	PAIS	Substitut.		867	2599	p.(Val867 Leu)				high			Male	Ambiguous	pos	Hiort et al. J Clin Endocrinol Metab, 77: 262-266, 1993
		7 LBD		3714	c.2599G	3714G >T										
0262	PAIS	Substitut.	*	867	2599	p.(Val867 Leu)			zero							Merkabi et al. 75th US Endo Soc Meeting Abstr 602, 1993
		7 LBD		3714	c.2599G	3714G >T										
0263	CAIS	Substitut.	**	867	2599	p.(Val867 Met)	20	16	normal	high			Female	Normal		Kazemi-Esfarjani et al. Mol Endocrinol, 7: 37 -46, 1993
		7 LBD		3714	c.2599G	3714G >A										
0264	CAIS	Substitut.	**	867	2599	p.(Val867 Met)			normal	high			Female	Normal		Weidemann et al. Clin Endocrinology 45: 733 -
		7 LBD		3714	c.2599G	3714G >A										
0265	CAIS	Substitut.	*	867	2599	p.(Val867 Met)			normal	high	*		Female	Normal		Lubahn et al. Proc Natl Acad Sci. 86: 9534, 1989
		7 LBD		3714	c.2599G	3714G >A										
0266	PAIS	Substitut.	*	867	2599	p.(Val867 Met)					*					McPhaul et al. J Clin Inv, 90: 2097, 1992
		7 LBD		3714	c.2599G	3714G >A										
0267	PAIS	Substitut.	*	867	2599	p.(Val867 Met)				high	*	de novo mutation - mosaic 2 functionally diff AR's	Female	Ambiguous	neg	Hiort et al. J Pediatrics 132: 939- 943, 1998
		7 LBD		3714	c.2599G	3714G >A										
0373	Prostate cancer	Substitut.	*	867	2599	p.(Val867 Met)						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
		7 LBD		3714	c.2599G	3714G >A										
0473	CAIS	Substitut.	*	867	2599	p.(Val867 Met)						Vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		7 LBD		3714	c.2599G	3714G >A										
0474	CAIS	Substitut.	*	867	2599	p.(Val867 Met)			zero			Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		7 LBD		3714	c.2599G	3714G >A										
0475	CAIS	Substitut.	*	867	2599	p.(Val867 Met)			zero			Epididymis present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
		7 LBD		3714	c.2599G	3714G >A										
0607	CAIS	Substitut.	*	867	2599	p.(Val867 Met)			zero				Female	Normal		Holterhaus et al. Genome Biology 4: R37
		7 LBD		3714	c.2599G	3714G >A										
0738	PAIS	Substitut.	*	867	2599	p.(Val867 Met)							Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
		7 LBD		3714	c.2599G	3714G >A										
0739	CAIS	Substitut.	*	867	2599	p.(Val867 Met)							Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
		7 LBD		3714	c.2599G	3714G >A										
0574	Prostate Cancer	Substitut.	*	867	2599	p.(Val867 Met)						Somatic mutation. + orchiectomy treatment Horm-	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
		7 LBD		3714	c.2599G	3714G >A										
0778	MAIS	Substitut.	*	867	2599	p.(Val867 Met)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606 -610, 2006
		7 LBD		3714	c.2599G	3714G >A										
0268	CAIS	Substitut.		867	2600	p.(Val867 Glu)							Female	Normal		McPhaul et al. J Clin Inv, 90: 2097, 1992
		7 LBD		3715	c.2600T	3715T >A										

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0855	CAIS	7 LBD		867		p.(Val867 fs)						Frameshift & stop in Codon 876 Diag 15yr Bilat gonadect at 16y	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0856	CAIS	7 LBD		867		p.(Val867 fs)						Frameshift & stop in Codon 876 Diag & Bilat gonadect at 11yr	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0259	PAIS	Substitut. 7 LBD		867 3714	2599 c.2599G	p.(Val867 Leu) 3714G >T	21		normal	high			Male	Ambiguous	pos	Saunders et al. Clin Endocrinol 37: 214, 1992
0851	CAIS	Substitut. 7 LBD		868 3717	2602 c.2602C	p.(Gln868 *) 3717C >T						Diag at 2.5 yr	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0852	CAIS	Substitut. 7 LBD		868 3717	2602 c.2602C	p.(Gln868 *) 3717C >T						Diag at 2 wk	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0940	Prostate Cancer	Substitut. 7 LBD		868 3717	2602 c.2602C	p.(Gln868 *) 3717C >T						Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
1054	Female Breast Caner	Substitut. 7 LBD		868 3719	2604 c.2604G	p.(Gln868 His 3719G >T						MDA-MB-453 breast cancer cell line reduced sensi to DHT	Female	Normal		Moore et al. Endocr Related Cancer 19:599 -613, 2012
0601	CAIS	Insertion 7 LBD		869		p.(Pro869) insATG						3 base insertion (ATG) at codon 869	Female	Normal		Scheiber et al. J Pediatric Endo Metab 16: 367-373, 2003
0269	PAIS	Substitut. 8 LBD	*	870 3725	2610 c.2610T	p.(Ile870 Met) 3725T >G			normal	high		* Hypospadia	Male	Ambiguous	pos	Bevan et al. Hum Mol Genet 5: 265-273, 1996
0718	PAIS	Substitut. 8 LBD		870 3725	2610 c.2610T	p.(Ile870 Met) 3725T >G				high			Male	Ambiguous	pos	Deeb et al. Clinical Endocrinology 63: 56 -62, 2005
0270	PAIS	Substitut. 8 LBD	*	871 3727	2612 c.2612C	p.(Ala871 Val) 3727C <T						Found in two unrelated families	Male	Ambiguous		Hiort et al. Eur J Pediatr, 153: 317, 1994
0315	PAIS	Substitut. 8 LBD		871 3727	2612 c.2612C	p.(Ala871 Gly) 3727C >G						Servere hypospadias	Male	Ambiguous		Albers et al. J of Pediatrics 131: 388 -392, 1997
0271	PAIS	Substitut. 8 LBD	*	871 3727	2612 c.2612C	p.(Ala871 Gly) 3727C >G						de novo mutation	Female	Ambiguous	neg	Hiort et al. J Pediatrics 132: 939- 943, 1998
0562	MAIS	Substitut. 8 LBD	*	871 3727	2612 c.2612C	p.(Ala871 Gly) 3727C >G						bilateral gynecomastia normal fertility	Male	Normal		Zenteno et al. Horm Res 57: 90-93, 2002
0794	CAIS ?	Deletion 8 LBD		871 3728	2613 c.2613	p.(Ala871) 3728 delG						1 nt deletion				Mueller et al. Hum Genet 119: 673, 2006
0951	PAIS	Substitut. 8 LBD	*	871 3727	2612 c.2612C	p.(Ala871 Val) 3727C >T						Micropenis only- Hetrozygous for SRD5A2 V89L	Male	Ambiguous		Bhangoo et al. Asian J Androl 12: 561-566, 2010
1010	PAIS	Substitut. 8 LBD	*	871 3727	2612 c.2612C	p.(Ala871 Val) 3727C >T	25	11					Male	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1098	PAIS	Substitut. 8 LBD	*	871 3727	2612 c.2612C	p.(Ala871 Val) 3727C >T										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
1099	PAIS	Substitut. 8 LBD	*	871 3727	2612 c.2612C	p.(Ala871 Glu) 3727C >A										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0272	MAIS	Substitut. 8 LBD	*	872 3729	2614 c.2614A	p.(Arg872 Gly) 3729A >G	26	24	normal	normal	norm	Gynocomastia & oligospermia	Male	Normal		Shkolny et al. J Clin Endocrinol Metab 84: 805-810, 1999
0696	Prostate cancer	Substitut. 8 LBD	*	873 3732	2617 c.2617G	p.(Glu873 Gln) 3732G >C	24		normal	normal		Activated by estrodiol, progesterone & CPA	Male	Normal		Chen et al. The Prostate 63: 395-406, 2005
0273	Prostate cancer	Substitut. 8 LBD		875 3738	2623 c.2623C	p.(His875 Tyr) 3738C >T						Som mut- stimulated by progesterone & oestrogen	Male	Normal		Taplin et al. New England J Med 332: 1393-1398, 1995
0274	Prostate cancer	Substitut. 8 LBD		875 3738	2623 c.2623C	p.(His875 Tyr) 3738C >T						Somatic mutation	Male	Normal		Tan et al. J of Urology 155: 340A, 1996
0538	CAIS	Substitut. 8 LBD		875 3739	2624 c.2624A	p.(His875 Arg) 3739A >G			zero				Female	Normal		Chavez et al. J Hum Genet. 46: 560-565, 2001
0868	CAIS	Substitut. 8 LBD		875 3739	2624 c.2624A	p.(His875 Arg) 3739A >G						Prenatal diagnosis	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008

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0869	CAIS	Substitut.	8 LBD	875	2624	p.(His875 Arg)						Prenatal diagnosis	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
				3739	c.2624A	3739A >G										
0870	CAIS	Substitut.	8 LBD	875	2624	p.(His875 Arg)						Diagnosis at 2mo bilateral gnadectomy at 16yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
				3739	c.2624A	3739A >G										
0871	CAIS	Substitut.	8 LBD	875	2624	p.(His875 Arg)						Diagnosis at 1yr bilateral gonadectomy at 15yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
				3739	c.2624A	3739A >G										
0872	CAIS	Substitut.	8 LBD	875	2624	p.(His875 Arg)						Diagnosis at 7 yrs bilateral gonadectomy at 14yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
				3739	c.2624A	3739A >G										
0873	CAIS	Substitut.	8 LBD	875	2624	p.(His875 Arg)						Prenatal diagnosis & bilateral gonadectomy at 14yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
				3739	c.2624A	3739A >G										
0275	LNCaP cell line	Substitut.	8 LBD	878	2632	p.(Thr878 Ala)						Altered binding specificity - somatic mutation	Male	Normal		Veldscholte et al. Biochem Biophys Res Comm, 172: 534, 1990
				3747	c.2624A	3747A >G										
0276	Prostate cancer	Substitut.	8 LBD	878	2632	p.(Thr878 Ala)						Somatic mutation 1/8 endocrine resistant therapy cases	Male	Normal		Suzuki et al. J Steroid Biochem Molec Biol 46: 759, 1993
				3747	c.2624A	3747A >G										
0277	Prostate cancer	Substitut.	8 LBD	878	2632	p.(Thr878 Ala)						6 out of 24 patients screened - somatic mutation	Male	Normal		Gaddipati et al. Cancer Res, 54: 2861-2864, 1994
				3747	c.2624A	3747A >G										
0278	Prostate cancer	Substitut.	8 LBD	878	2632	p.(Thr878 Ala)						3 out of 22 cases in metastatic tissue - somatic mutation	Male	Normal		Suzuki et al. Prostate 29: 153-158, 1996
				3747	c.2624A	3747A >G										
0279	Prostate cancer	Substitut.	8 LBD	878	2632	p.(Thr878 Ala)						Somatic mutation in bone metasteses of Prostate cancer	Male	Normal		Kleinerman et al. J of Urology 155: 624A, 1996
				3747	c.2624A	3747A >G										
0432	Prostate cancer	Substitut.	8 LBD	878	2632	p.(Thr878 Ala)						Som mut found in 5 of 16 patients treated with flutamide	Male	Normal		Taplin et al. Cancer Research 59: 2511 -2515
				3747	c.2624A	3747A >G										
0603	Prostate cancer	Substitut.	8 LBD	878	2632	p.(Thr878 Ala)						Somatic mutation - flutamide treated	Male	Normal		Taplin et al. J Clinical Oncology 21: 2673 -2678, 2003
				3747	c.2624A	3747A >G										
0604	Prostate cancer	Substitut.	8 LBD	878	2632	p.(Thr878 Ala)						Somatic mutation - flutamide treated	Male	Normal		Taplin et al. J Clinical Oncology 21: 2673 -2678, 2003
				3747	c.2624A	3747A >G										
0647	Prostate cancer	Substitut.	8 LBD *	878	2632	p.(Thr878 Ala)						+Q641*- late stage disease- som mut - transactivates	Male	Normal		Ceraline et al. Intl J Cancer 108: 152-157, 2003
				3747	c.2624A	3747A >G										
0280	Prostate cancer	Substitut.	8 LBD *	878	2633	p.(Thr878 Ser)						Som mut. in 86% of isolates.Stimulated by estrogen & progester	Male	Normal		Taplin et al: New England J Med 332: 1393-1398, 1995
				3748	c.2633C	3748C >G										
1026	AIS	Substitut.	8 LBD	878	2633	p.(Thr878 Ile)										Mueller et al. Hum Gen 127: 479, 2010
				3748	c.2633C	3748C >T										
0539	PAIS	Substitut.	8 LBD	880	2638	p.(Asp880 Tyr)			normal				Male	Ambiguous		Chavez et al. J Hum Genet. 46: 560-565, 2001
				3753	c.2638G	3753G >T										
0553	Prostate cancer	Substitut.	8 LBD	880	2639	p.(Asp880 Gly)						Treated with bicalumatide - somatic mutatation	Male	Normal		Taplin et al. J Clinical Oncology 21: 2673 -2678, 2003
				3754	c.2639A	3754A >C										
0281	CAIS	Substitut.	8 LBD	882	2644	p.(Leu882 Val)						Somatic instabilty in polyglutamine tract	Female	Normal	pos	Davies et al. Clinical Endocrinology 43: 69 -77, 1995
				3759	c.2644C	3759C >G										
1100	PAIS	Substitut.	8 LBD	882	2644	p.(Leu882 Ile)										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
				3759	c.2644C	3759C >A										
0829	CAIS	Substitut.	8 LBD	882	2645	p.(Leu882 Pro)							Female	Normal	neg	Galani et al. Fertility & Sterility 2008
				3760	c.2645T	3760T >C										
0282	CAIS	Substitut.	8 LBD	884	2650	p.(Lys884 *)			zero				Female	Normal	pos	Trifiro et al. Am J Med Genet, 40: 493, 1991
				3765	c.2650A	3765A >T										
0623	CAIS	Substitut.	8 LBD	885	2654	p.(Ser885 *)	23		v low				Female	Normal		MacLean et al. Hum Mutat. 23: 287, 2004
				3769	c.2654C	3769C >G										
0283	MAIS	Substitut.	8 LBD *	887	2659	p.(Met887 Val)	23	23	normal	normal	norm	Oligospermia-50% red. in transactivation	Male	Normal		Ghadessy et al. J. Clin. Endocrinol. 103: 1517 -1525, 1999
				3774	c.2659A	3774A >G										
0309	MAIS	Substitut.	8 LBD *	887	2659	p.(Met887 Val)	21	24	normal	normal	norm	Oligospermia-50% red. in transactivation	Male	Normal		Ghadessy et al. J. Clin. Endocrinol. 103: 1517 -1525, 1999
				3774	c.2659A	3774A >G										

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0803	Liver cancer	Substitut. 8 LBD	*	887 3774	2659 c.2659A	p.(Met887 Val) 3774A >G	21	24				Som mut ? + T1034G in 5'UTR. found in tum & non-tumorous	Male	Normal		Yeh et al. Int J Cancer 120: 1610-1617, 2007
0697	Prostate cancer	Substitut. 8 LBD	*	887 3776	2661 c.2661G	p.(Met887 Ile) 3776G >A	21		normal	normal		Increased response to TIF-2 & CBP w DHT w N-CoR > inhibition	Male	Normal		Chen et al. The Prostate 63: 395-406, 2005
0593	PAIS	Substitut. 8 LBD		*889 3782	2667 c.2667_2668	p.(Ser889 Lys) 3782_3783 delins AG						no immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87. 182-188. 2002
0533	PAIS	Substitut. / Splice 8 LBD		*889 3782	2667 c.2667C	p.(Ser889) 3782C >T	21	24	v low	normal		silent mut.-part exon 8+ part of 3' untransl also small amt. wt AR	Male	Ambiguous		Hellwinkel et al. J Clin Endocrinol Metab 86: 2569-2575, 2001
0540	PAIS	Substitut/ Splice 8 LBD		*889 3782	2667 c.2667C	p.(Ser889) 3782C >T			normal			silent mutation	Male	Ambiguous		Chavez et al. JHum Genet. 46: 560-565 2001
1011	PAIS	Substitut. 8 LBD		*889 3782	2667 c.2667C	p.(Ser889) 3782C >T	23	17	zero	zero		mother heterozygote carrier	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1012	PAIS	Substitut. 8 LBD		*889 3782	2667 c.2667C	p.(Ser889) 3782C >T	25	18					Female	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0284	CAIS	Substitut. 8 LBD		*890 3783	2668 c.2668G	p.(Val890 Met) 3783G >A			zero				Female	Normal		Pinsky et al. Clin Inv Med, 15: 456, 1992
0285	PAIS	Substitut. 8 LBD	**	890 3783	2668 c.2668G	p.(Va890 Met) 3783G >A			low	normal			Female	Ambiguous		De Bellis et al. J Clin Endocrinol Metab, 78: 513, 1994
0321	PAIS	Substitut. 8 LBD		*890 3783	2668 c.2668G	p.(Val890 Met) 3783G >A							Female	Normal		Essawi et al. Disease Markers 13: 99-105, 1997
0476	CAIS	Substitut. 8 LBD		*890 3783	2668 c.2668G	p.(Val890 Met) 3783G >A			low	normal			Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0624	CAIS	Substitut. 8 LBD		*890 3783	2668 c.2668G	p.(Val890 Met) 3783G >A	22		low				Female	Normal		MacLean et al. Hum Mutat. 23: 287, 2004
0740	CAIS	Substitut. 8 LBD		*890 3783	2668 c.2668G	p.(Val890 Met) 3783G >A							Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
0999	CAIS	Substitut. 8 LBD		*890 3783	2668 c.2668G	p.(Val890 Met) 3783G >A	21	17				1 sister affected; mother heterozygote; 2 aunts wt	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1013	PAIS	Substitut. 8 LBD		*890 3783	2668 c.2668G	p.(Val890 Met) 3783G >A	18	14					Male	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0433	Prostate cancer	Substitut. 8 LBD		*891 3786	2671 c.2671G	p.(Asp891 Asn) 3786G >A						Mutation also found in peripheral blood	Male	Normal		Taplin et al. Cancer Research 59: 2511 -2515, 1999
1102	CAIS	Substitut. 8 LBD		893 3791	2676 c.2676C	p.(Pro893 Thr 3791C >A							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0389	CAIS	Substitut. 8 LBD	*	893 3792	2677 c.2677C	p.(Pro893 Ser) 3792C >T	26		low	high		Reduced transactivation	Female	Normal	neg	Peters et al. Mol & Cellular Endocrinol. 148: 47-53, 1999
0375	CAIS	Substitut. 8 LBD		893 3793	2678 c.2678C	p.(Pro893 Leu) 3793C >T						Mutation found in two siblings	Female	Normal	pos	Knoke et al. Human Mutation 12: 220, 1998
0413	CAIS	Substitut. 8 LBD		893 3793	2678 c.2678C	p.(Pro893 Leu) 3793C >T							Female	Normal		Kanayama et al. Int J Urology 6: 327-330, 1999
0741	CAIS	Substitut. 8 LBD		893 3793	2678 c.2678C	p.(Pro893 Leu) 3793C >T							Female	Normal		Ledig et al: Horm Res 63: 263-269, 2005
0742	CAIS	Substitut. 8 LBD		893 3793	2678 c.2678C	p.(Pro893 Leu) 3793C >T							Female	Normal		Ledig et al: Horm Res 63: 263-269, 2005
1101	CAIS	Substitut. 8 LBD		893 3793	2678 c.2678C	p.(Pro893 Leu) 3793C >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0795	CAIS	Deletion 8 LBD		895 3798	2683 c.2683-2685	p.(Met895 del) 3798-3800 delATG						3 nt deletion	Female	Normal		Mueller et al. Hum Genet 119: 672, 2006
0766	Prostate cancer	Substitut. 8 LBD		896 3801	2686 c.2686A	p.(Met896 Val) 3801A >G						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006

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0669	CAIS	Substitut.	8 LBD	896	2687	p.(Met896 Thr)						Somatic mosaicism	Male	Ambiguous		Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
				3802	c.2687T	3802T >C										
0386	CAIS	Substitut.	8 LBD *	896	2687	p.(Met896 Thr)			low			Reduced transactivation	Female	Normal		Giwercman et al. Human Genetics 103: 529-531, 1998
				3802	c.2687T	3802T >C										
1000	CAIS	Substitut.	8 LBD	896	2687	p.(Met896 Thr)	26	17					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
				3802	c.2687T	3802T >C										
1103	CAIS	Substitut.	8 LBD *	896	2687	p.(Met896 Thr)							Female	Normal		Hughes et al. Semin Reprod Med 30: 432 -442, 2012
				3802	c.2687T	3802T >C										
1104	PAIS	Substitut.	8 LBD	897	2690	p.(Ala897 Glu)										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
				3085	c.2690	3085C >A										
1105	PAIS	Substitut.	8 LBD	898	2693	p.898Glu Gly										Hughes et al. Semin Reprod Med 30: 432 -442, 2012
				3088	c.2693	3088A >G										
0642	PAIS	Substitut.	8 LBD	899	2695	p.(Ile899 Phe)	19					Testis located in labia majora- Separated vaginal & urethra opn	Female	Ambiguous	pos	Melo et al. J Clin Endocrinol Metab 88: 3241-3250, 2003
				3810	c.2695A	3810A >T										
0643	PAIS	Substitut.	8 LBD	899	2695	p.(Ile899 Phe)	19					Testis located in inguinal -Separated vaginal & urethra opn	Female	Ambiguous	pos	Melo et al. J Clin Endocrinol Metab 88: 3241-3250, 2003
				3810	c.2695A	3810A >T										
0644	PAIS	Substitut.	8 LBD	899	2695	p.(Ile899 Phe)	19					Testis located in inguinal- Single vaginal & urethra opn	Female	Ambiguous	pos	Melo et al. J Clin Endocrinol Metab 88: 3241-3250, 2003
				3810	c.2695A	3810A >T										
0645	PAIS	Substitut.	8 LBD	899	2695	p.(Ile899 Phe)	19					Testis located in inguinal -Separated vaginal & urethra opn	Female	Ambiguous	pos	Melo et al. J Clin Endocrinol Metab 88: 3241-3250, 2003
				3810	c.2695A	3810A >T										
0286	CAIS	Substitut.	8 LBD	899	2696	p.(Ile899 Thr)						de novo mutation	Female	Normal	neg	Hiort et al. J Pediatrics 132: 939- 943, 1998
				3811	c.2696T	3811T >C										
0649	PAIS	Substitut.	8 LBD *	903	2707	p.(Gln903 Lys)				high	high	Involved in TIF2 N/C interaction	Male	Ambiguous	pos	Umar et al. J Cin Endocrinol & Metab 90: 507-515, 2005
				3822	c.2707C	3822C >A										
1014	PAIS	Substitut.	8 LBD	903	2707	p.(Gln903 Lys)	21	17				mother heterozygote carrier	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
				3822	c.2707C	3822C >A										
0287	Prostate cancer	Substitut.	8 LBD	903	2708	p.(Gln903 Arg)						Somatic mutation in 37% of isolates in initial cloning	Male	Normal		Taplin et al. New England J Med 332: 1393-1398, 1995
				3823	c.2708A	3823A >G										
0288	PAIS	Substitut.	8 LBD	904	2710	p.(Val904 Met)			low			Qualitative binding abnormality				McPhaul et al. J Clin Inv 90: 2097, 1992
				3825	c.2710G	3825G >A										
1001	CAIS	Substitut.	8 LBD	904	2710	p.(Val904 Leu)	23	18				Mother heterozygote carrier	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
				3825	c.2710G	3825G >T										
0289	CAIS	Substitut.	8 LBD	905	2713	p.(Pro905 Ser)	27	23	normal	high			Female	Normal		Pinsky et al. Clin Inv Med, 15: 456-472, 1992
				3828	c.2713C	3828C >T										
0290	CAIS	Substitut.	8 LBD	905	2714	p.(Pro905 His)			zero				Female	Normal		McPhaul et al. J Clin Inv, 90: 2097, 1992
				3829	c.2714C	3829C >A										
0646	CAIS	Substitut.	8 LBD	905	2714	p.(Pro905 Val)	21					Testis located in inguinal region	Female	Normal	neg	Melo et al. J Clin Endocrinol Metab 88: 3241-3250, 2003
				3829	c.2714C	3829C >G										
1002	CAIS	Deletion	8 LBD	906	2715	p.(Pro905 fs)	22	17				mother heterozygote carrier	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
				3830	c.2715	3830 delC										
0291	CAIS	Substitut.	8 LBD *	908	2722	p.(Leu908 Phe)			low	normal		Decreased transactivat Epididymis & Vas	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
				3837	c.2722C	3837C >T										
0292	PAIS	Substitut.	8 LBD *	910	2728	p.(Gly910 Arg)			low	low		Also silent G to A mutation in codon 213	Female	Ambiguous	pos	Choong et al. J Clin Endocrinol Metab, 81: 236-243, 1996
				3843	c.2728G	3843G >A										
0374	Prostate cancer	Substitut.	8 LBD	910	2729	p.(Gly910 Glu)						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621 -1624, 1995
				3844	c.2729G	3844G >A										
0327	Prostate cancer	Substitut.	8 LBD	911	2732	p.(Lys911 Arg)						Somatic mutation	Male	Normal		Watanabe et al. Jpn J Clin Oncol 27: 389 -393, 1997
				3847	c.2732A	3847A >G										
0430	PAIS	Substitut.	8 LBD	912	2734	p.(Val912 Leu)	19					Servere oligozoospermia	Male	Ambiguous		Knoke et al. Andrologia 31: 199-201, 1999
				3849	c.2734G	3849G >C										

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base	Position: HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0293	PAIS	Substitut.		914	2740	p.(Pro914 Ser)							Female	Ambiguous		Ghirri and Brown. Paed Res, 33(5) Suppl, Abstr 95, 1993
		8 LBD		3855	c.2740C	3855C >T										
1015	PAIS	Substitut.		914	2740	p.(Pro914 Ser)			zero	zero			Female	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		8 LBD		3855	c.2740C	3855C >T										
0958	CAIS	Substitut.		914	2741	p.(Pro914 Arg)							Female	Normal	neg	Wu et al. Fertility & Sterility 93: 2076, e1-4, 2010
		8 LBD		3856	c.2741C	3856C >G										
0844	CAIS	Deletion	*	916		p.(Phe917 *)						12nt del. Stop in codon 917 affect sister & aunts. neg. N/C interaction	Female	Normal	pos	Werner et al. Sex Dev 2: 73-83, 2008
		8 LBD														
0318	CAIS	Substitut.	*	917	2751	p.(Phe917 Leu)			low	high	*		Female	Normal		Radnayr et al. J of Urology 158: 1553 -1556, 1997
		8 LBD		3866	c.3866C	3866C >G										
0477	CAIS	Substitut.		918	2753	p.(His918 Arg)							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		8 LBD		3868	c.3868A	3868A >G										
0303	Prostate cancer	Substitut.	*	920	2759	p.(Gln920 Arg)						Somatic mutation	Male	Normal		Nazareth et al. 79th US Endo Soc Meetings Abstr. P2-498, 1997
				3874	c.3874A	3874 >G										
0294	CAIS	Splice					24	23				Insertion at +3 position of donor splice site	Female	Normal		Trifiro et al. Eur J Hum Genetics 5: 50-58, 1997
		exon1/ intron 1			c.1616+2 1616+3	2731+2_2731 +3 insT										
0906	CAIS	Splice										Substitution at +5 of donor spice site	Female	Normal	pos	Philibert et al. Fertility & Sterility 94: 472-476 2010
		exon1/ intron 1			c.1616+5G	2731+5G >C										
0304	CAIS	Splice										Substitution at +1 pos of donor splice site - lacks exon 2	Female	Normal	neg	Hellwinkel et al. J Steroid Biochem Mol Biol 68: 1-9, 1999
		exon2/ intron 2			c.1768+1G	2883+1G >A										
0479	CAIS	Splice							zero				Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		exon2/ intron 2														
0480	CAIS	Splice											Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		exon2/ intron 2														
0351	CAIS	Splice										Substition at + 1 pos of donor splice site	Female	Normal		Hiort et al. J Pediatrics 132: 939- 943, 1998
		exon2/ intron 2			c.1768+1G	2883+1G >A										
0317	Breast Cancer	Deletion										delta-exon3AR mRNA: higher express of mut. var in 7/13 breast cancer	Female	Normal		Zhu X et al. Int J Cancer 72: 574-580, 1997
		exon3														
1022	Premature ovarian failure	Substitut.										Substitution at +9 of donor site. Patient had menopause at 18	Female	Normal		Panda et al. Gyncol Endocrinol 27: 1-7, 2011
		exon3/ intron 3			c.1885+9C	3000+9C >A										
0295	CAIS	Splice										Substitution at +1 position of donor splice site	Female	Normal		Evans et al. J Endocrinol 129 Suppl, Abstr 65, 1991
		exon3/ intron 3			c.1885+1G	3000+1G >A										
0478	CAIS	Splice							normal	normal		Substitution at +1 position of donor splice site	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
		exon3/ intron 3			c.1885+1G	3000+1G >A										
0296	CAIS	Splice							zero			+1 pos of donor site. Splice site activated & p.(684G_724Pdel)	Female	Normal		Ris-Stalpers et al. Proc Natl AcadSci 87: 7866 -70, 1990
		exon4/ intron 4			c.2173+1G	3288+1G >T										
1057	CAIS	Splice										Mother + a sister carriers	Female	Normal	pos	Vasu et al. Genet Test Mol Biomarkers 16:749 -755, 2012
		exon5/ intron 5			c.2320+1G	3435+1G >A										
1018	CAIS	Splice					28	17	low	normal		Substit. at -44 pos of donor acceptor site	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
		exon6/ intron 6			c.2450-44G	3565-44G >A										
0503	PAIS	Splice							low	normal		Substit. at +5 position of donor splice site, stop at + 79 bases	Female	Ambiguous		Sammarco et al. J Clin Endocrinol Metab 85: 3256-3261, 2000
		exon6/ intron 6			c.2449+5G	3564+5G >T										
0541	CAIS	Splice							zero			Substit. at +5 position of donor splice site.	Female	Normal		Chavez et al. J Hum Genet 46: 560-565, 2001
		exon6/ intron 6			c.2449+5G	3564+5G >C										
1029	CAIS	Splice										Substitution at +3 pos of donor splice site. Exon 6 skipped				Trifiro et al. Eur J Hum Genetics 5: 50-58, 1997
		exon6/ intron 6			c.2449+3A	3564+3A >T										
0298	CAIS	Splice							zero			Substit. +1 pos donor splice, mRNA exon 7del, stop + 10 aa from exon 8	Female	Normal	pos	Lim et al. Mol & Cell Endocrinology 131: 205 -210, 1997
		exon7/ intron 7			c.2607+1G	3722+1G >A										
0502	CAIS	Splice										Substitution at + 1 position of donor splice site	Female	Normal		Choi et al. Arch Gynecol Obstet 263: 201-205, 2000
		exon7/ intron 7			c.2607+1G	3722+1G >T										

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position: Amino acid / Base / HGVS nt / HGVS base	Change: Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0670	CAIS	Splice	exon7/ intron 7		c.2607+1G	3722+1G >A				normal	normal	Substit. at + 1 pos of donor splice site somtaic mosaicsm	Female	Normal		Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
0088	PAIS	Deletion	intron 2		c.?_1769-19	?_2884-19 del				normal	normal	≥6 kb del at -19 pos of acceptor site. 2 transcr: wt, exon3del	Male	Ambiguous	pos	Ris-Stalpers et al. Am J Hum Genet 54: 609, 1994
0749	PAIS	Deletion	intron 2							normal	high	6 kb del of intron 2, affects splicing, related to 0750	Male	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0750	PAIS	Deletion	intron 2									related to 0749	Male	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0299	PAIS	Splice	intron 2/exon 3		c.1769-11T	2884-11T >A						Subst. -11 slice accept. 2 transc: exon3del & +69 nt; 3 affct sib, var phen	Male	Normal	pos	Bruggenwirth et al. Am J Hum Genet 61: 1067-1077, 1997
0809	Prostate cancer	Splice	intron 2/exon 3	*	c.1768_1769	2883_2884 ins69						AR23 variant, ins 69 nt of intron 2 = 23 aa, affects AR trafficking	Male	Normal		Jagla et al. Endocrinology 148: 4334-43, 2007
1017	CAIS	Splice	intron 2/exon 3		c.1769-3C	2884-3C >G	26	18				Substit at -3 of acceptor site. Aborted fetus	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0909	Prostate cancer	Splice	intron 2/exon 3		c.1768_1769	2883_2884 ins69						AR23 splice variant, ins 69 nt of intron 2, found in 5/8 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0800	CAIS	Splice	intron 2/exon 3		c.1769-1G	2884-1G >A						Subst. -1 pos accept. transc: 2883_2884ins69	Male	Normal		Jaaskelainen et al. Hum Mutat 27: 291, 2006
1016	CAIS	Splice	intron 2/exon 3		c.1769-2A	2884-2A >C	13	18				Substit at -2 of acceptor site. Also Lys591Glu mut	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0594	CAIS	Splice	intron 3/exon 4		c.1886-1G	3001-1G >T						Substit at -1 of acceptor site.	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0835	PAIS	Substitut.	intron 3/exon 4		c.1886-60G	3001-60G >A						Male to female	Female	Ambiguous		Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
0323	Prostate cancer		3'UTR									Polymorphism in 3"UTR Somatic mutation	Male	Normal		Paz et al. European Urology 63:56-62, 2005