March 2012

Syndrome of Hepatic Cirrhosis, Dystonia, Polycythemia, and Hypermanganesemia Caused by Mutations in SLC30A10, a Manganese Transporter in Man

Karin Tuschl

Peter T. Clayton

Sidney M., Jr. Gospe

Shamshad Gulab
Aga Khan University

Shahnaz Ibrahim
Aga Khan University

See next page for additional authors

Authors
Karin Tuschl; Peter T. Clayton; Sidney M., Jr. Gospe; Shamshad Gulab; Shahnaz Ibrahim; Pratibha Singhi; Roosy Aulakh; Reinaldo T. Ribeiro; Orlando G. Barsottini; Maha S. Zaki; Maria Luz Del Rosario; Sarah Dyack; Victoria Price; andrea Rideout; Kevin Gordon; Ron A. Wevers; W. K. 'Kling' Chong; and Philippa B. Mills

Syndrome of Hepatic Cirrhosis, Dystonia, Polycythemia, and Hypermanganesemia Caused by Mutations in *SLC30A10*, a Manganese Transporter in Man

Karin Tuschl,* Peter T. Clayton, Sidney M. Gospe Jr., Shamshad Gulab, Shahnaz Ibrahim, Pratibha Singhi, Roosy Aulakh, Reinaldo T. Ribeiro, Orlando G. Barsottini, Maha S. Zaki, Maria Luz Del Rosario, Sarah Dyack, Victoria Price, Andrea Rideout, Kevin Gordon, Ron A. Wevers, W.K. ''Kling'' Chong, and Philippa B. Mills

(The American Journal of Human Genetics *90*, 457–466; March 9, 2012)

In this article, Figure 1 contained an error regarding the position of the deletion detected in the affected individuals of family A. The no-call SNPs between the coordinates 218,057,426 and 218,158,564 (GRCh36) suggest that exons 3 and 4 of *SLC30A10* are deleted, whereas the original figure showed that exons 1 and 2 were deleted. The corrected Figure 1 and its legend are shown here. The authors regret the error.

A

Subject	Exon	Mutation	
A1-A4	3-4	g.1qdel218.057.426_218.158.564bp	
B5-B7	1	c.266T>C,	p.Leu89Pro
C8	1	c.292_402del,	p.Val98_Phe134del
D9*	1	c.314_322del	p.Ala105_Pro107del
E11	1	c.585del	p.Thr196Profs*17
F12	3	c.765_767del	p.Val256del
G13*	3	c.922C>T	p.Gln308*
H15	4	c.1046T>C	p.Leu349Pro

B

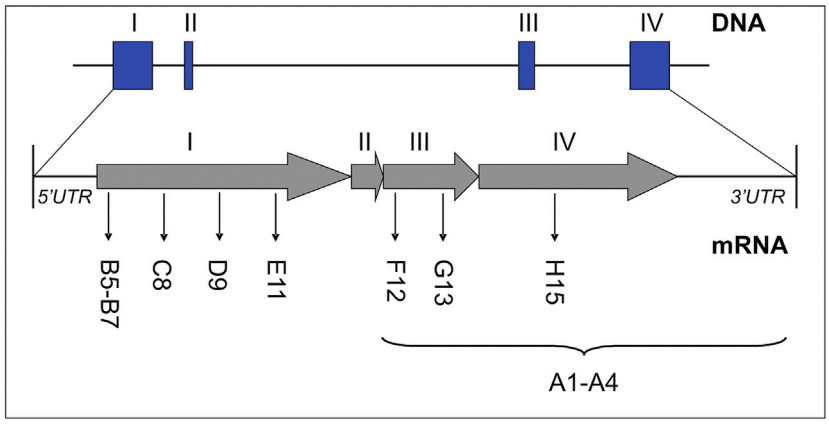

Figure 1. *SLC30A10* Mutations in Affected Families with a Syndrome of Hepatic Cirrhosis, Dystonia, Polycythemia, and Hypermanganesemia

(A) Mutations in *SLC30A10* identified by DNA sequencing. (For families D and G no DNA was available for analysis of deceased siblings D-II-3 and G-II-1.)

(B) Genomic structure of the exons encoding *SLC30A10* and positions of identified mutations. The large deletion spanning exons 3 and 4 in family A is indicated by a bracket.

*Correspondence: k.tuschl@ucl.ac.uk

http://dx.doi.org/10.1016/j.ajhg.2016.07.015.