

The London Conference
on the
Normal Human Karyotype¹

28th - 30th August, 1963

The present meeting was called to consider developments since the Denver Conference and to assess the degree to which these may have aided characterization of individual chromosomes or may have revealed inadequacies in the earlier identifications. There was general agreement that the seven groups proposed in the Denver numbering system have stood the test of time, and that little or no uncertainty exists in the group assignment of any chromosome. However within certain groups identification of individual chromosomes is questionable. The groups are sometimes designated by letters, such as A to G, as well as by the numbering scheme described in the Denver classification.

A comparison of data submitted by different investigators revealed that surprisingly good agreement on relative chromosomal length and arm ratios or centromeric indices can be obtained in different laboratories provided that mitotic figures, clearly delineated and free from irregularities of condensation pattern, are selected for analysis. The dispersion of chromosomal measurements appears to be due to both biological and technical factors and, while some of these variations are random, others appear to be systematically produced, as in the effect of exposure to colchicine which may contract some chromosomes more than others. It is hoped that specific procedures maximizing ease of identification will soon be forthcoming.

The new techniques considered included the demonstration of specific chromosomal constrictions and autoradiographic determination of the time at which H^3 thymidine is incorporated in specific

¹ Ciba Foundation Guest Symposium sponsored by The Association for the Aid of Crippled Children, New York.

chromosomes and chromosomal regions. Both of these methods contribute to the characterization of specific chromosomes. However variability exists in techniques and material; so that at present neither method permits absolute identification of chromosomes. There was discussion on the use of methods for DNA measurement in each chromosome arm, on electron microscopy, on satellite association, on specific location of certain chromosomes within spread mitotic figures, on physico-chemical separation of chromosomes in suspension, on the use of reagents which may characterize chromosome segments and on the ash patterns produced by micro-incineration.

It was agreed at Denver that the autosomes should be serially numbered 1 to 22 as nearly as possible in descending order of length. Identification of individual chromosomes is based on size, position of centromere and other morphological features. The sex chromosomes are referred to as X and Y.

The most important newly recognized features are so-called secondary constrictions¹ and differential patterns of incorporation of isotopically labelled thymidine in specific chromosomes as demonstrated by autoradiography. Secondary constrictions are only seen in certain types of preparations and then in a fraction of the cells.

Variability is observable in the morphology of certain chromosomes. This is especially well established for the Y chromosomes in which great differences in size occur and which are heritable from father to son. Heritable polymorphism has also been observed in satellited chromosomes and probably in No. 16.

Specific Comments

It is emphasized that assignment of numbers to chromosome pairs on the basis of the morphological criteria here discussed includes an element of arbitrariness; it does not ensure homology.

Group 1-3, (A)

No. 1. In many cells a secondary constriction is observed in the proximal region of the long arm.

Group 4-5, (B)

No additional information is available which can be used to distinguish these two chromosome pairs.

¹ Apart from satellite stalks the term 'secondary constriction' connotes achromatic regions which may appear as attenuated parts.

Group 6-12 and the X chromosome, (C)

Of the autosomes in this group, four are comparatively metacentric. It is proposed that they should be numbered 6, 7, 8 and 11. The X chromosome belongs to this sub-group.

Three chromosomes are sub-metacentric. They should be numbered 9, 10 and 12.

A secondary constriction is found in the proximal part of the long arm of at least one of the pairs in the whole group.

In normal female cells, one chromosome characteristically incorporates isotopically labelled thymidine over most of its length later than the others in the group. This chromosome is believed to be an X.

Group 13-15, (D)

Satellites have now been detected in all three chromosome pairs.

Group 16-18, (E)

A secondary constriction has been frequently seen in the proximal part of the long arm of No. 16.

Group 19-20, (F)

No additional information is available which can be used to distinguish these two chromosome pairs.

Group 21-22 and the Y chromosome, (G)

Satellites have now been detected on both Nos. 21 and 22.

The most common Y is larger than either 21 or 22, its centric constriction is often indistinct and a secondary constriction is frequently seen in the long arm; the terminal region of the long arm may be poorly defined. Typically, the two long arm chromatids of the Y appear to diverge less than those of other chromosomes.

Participants

N. A. BARNICOT,
Department of Anthropology,
University College London,
Gower Street,
London, W. C. 1 (England)

D. H. CARR,
Department of Microscopic
Anatomy,
University of Western Ontario,
346 South Street,
London, Ontario (Canada)

M. A. BENDER,
Biology Division,
Oak Ridge National Laboratory,
Post Office Box Y,
Oak Ridge, Tennessee (USA)

E. H. Y. CHU,
Biology Division,
Oak Ridge National Laboratory,
Post Office Box Y,
Oak Ridge, Tennessee (USA)

M. A. FERGUSON-SMITH,
Genetics Department,
The University,
Glasgow, W. 2 (Scotland)

J. LEJEUNE,
Institut de Progénèse,
15, rue de l'École de Médecine,
Paris VIe (France)

C. E. FORD,
M.R.C. Radiobiological
Research Unit, Harwell,
Didcot, Berks. (England)

W. LENZ,
Institut für Humangenetik der
Universität,
Universitäts-Krankenhaus
Eppendorf,
Martinstraße 52,
Hamburg 20 (Germany)

M. FRACCARO,
Gruppo Euratom i Università di
Pavia,
Via Forlanini, 14,
Pavia (Italy)

J. LINDSTEN,
Department of Endocrinology
and Metabolism,
Karolinska Sjukhuset,
Stockholm 60 (Sweden)

J. GERMAN,
The Rockefeller Institute,
New York, 21, N. Y. (USA)

J. L. HAMERTON,
Paediatric Research Unit,
Cameron House,
Guy's Hospital Medical School,
London, S. E. 1 (England)

O. J. MILLER,
Department of Obstetrics and
Gynaecology,
College of Physicians and
Surgeons of Columbia University,
New York 32, N. Y. (USA)

K. HIRSCHHORN,
Department of Medicine,
New York University School of
Medicine,
550 First Avenue,
New York 16, N. Y. (USA)

P. S. MOORHEAD,
Wistar Institute,
Thirty-Sixth Street at Spruce,
Philadelphia 4, Penn. (USA)

P. A. JACOBS,
M.R.C. Clinical Effects of
Radiation Research Unit,
Western General Hospital,
Crewe Road,
Edinburg 4 (Scotland)

K. PATAU,
University of Wisconsin,
Department of Medical Genetics,
Genetics Building,
Madison 6, Wisc. (USA)

L. S. PENROSE (convener),
Galton Laboratory,
University College London,
Gower Street,
London, W. C. 1 (England)

T. T. PUCK,
Department of Biophysics,
University of Colorado,
4200 East Ninth Avenue,
Denver 20, Colorado (USA)

A. ROBINSON,
Department of Biophysics,
University of Colorado,
4200 East Ninth Avenue,
Denver 20, Colorado (USA)

W. SCHMID,
M.D. Anderson Hospital and
Tumor Institute,
Texas Medical Center,
Houston 25, Texas (USA)
and
Kinderspital,
Zürich (Switzerland)

C. STERN,
Department of Zoology,
University of California,
Berkeley 4, Calif. (USA)

J. H. TJIO,
National Institutes of Health,
Bethesda 14, Md. (USA)

M. J. D. WHITE,
Department of Zoology,
The University,
Melbourne (Australia)