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In 1956 Tjio and Levan, in their now classic paper, reported that the human chromosome number was 46 and not 48. This work, which was carried out on cultured human embryonic cells, was rapidly confirmed by studies of testicular material by Ford and Hamerton (1956). These two papers stimulated a renewed interest in human cytogenetics, and, by 1959, several laboratories were engaged in the study of human chromosomes and a variety of classification and nomenclature systems had been proposed. This resulted in confusion in the literature and a need to establish a common system of nomenclature that would improve communication between workers in the field.

For this reason, a small study group was convened in Denver, Colorado, at the suggestion of Dr. Charles E. Ford. Fourteen investigators and three consultants participated, representing each of the laboratories that had published human karyotypes up to that time. The system proposed in the report of this meeting, entitled “A Proposed Standard System of Nomenclature of Human Mitotic Chromosomes,” more usually known as the Denver Conference (1960), has formed the basis for all subsequent nomenclature reports and has remained virtually unaltered, despite the rapid developments of the last 18 years. It is fair to say that the participants at Denver did their job so well that this report has formed the cornerstone of human cytogenetics since 1960, and the foresight and cooperation shown by these investigators have prevented much of the nomenclature confusion which has marked other areas of human genetics.
Three years later, a meeting called by Professor Lionel S. Penrose was held in London (London Conference, 1963) to consider developments since the Denver conference. The only significant result of that conference was to give official sanction to the classification of the seven groups of chromosomes by the letters A to G, as originally proposed by Patau (1960). When referring to this report in text, it should be cited as ISCN (1978). The complete citation for reference lists is: ISCN (1978): An International System for Human Cytogenetic Nomenclature (1978). Birth Defects: Original Article Series, Vol. XIV, No. 8 (The National Foundation, New York 1978); also in Cytogenet. Cell Genet. 21: 309-404 (1978).

The next significant development came in Chicago at the Third International Conference on Human Genetics in 1966 when 37 investigators, representing the major cytogenetic laboratories, met to determine whether it was possible to improve the nomenclature and thus eliminate some of the major problems that had resulted from the rapid proliferation of new findings since 1960. The report of this conference (Chicago Conference, 1966) proposed a standard system of nomenclature for the provision of short-hand descriptions of the human chromosome complement and its abnormalities, a system that, in its basic form, has stood the test of time and is now used throughout the world for the description of non-banded chromosomes.

In his introductory address to the Chicago Conference (1966), Lionel Penrose made the following prophetic statement:

“It is easy to be carried away by the detectable peculiarities and to forget that much underlying variability is still hidden from view until some new technical device discloses the finer structure of chromosomes, as in the Drosophila salivary gland cells.”

Two years later, in 1968, the second major breakthrough occurred when T. Caspersson and his colleagues, working in Sweden, published the first banding pictures of plant chromosomes stained with quinacrine dihydrochloride or quinacrine mustard (Caspersson et al., 1968). These studies were rapidly expanded to human chromosomes by these workers, who published the first banded human karyotype in 1970 (for a review of this work, see Caspersson et al., 1971). Soon, several other techniques that also produced chromosome bands were developed. This led to the realization that, as each human chromosome could now be identified very precisely, the existing system of nomenclature would no longer be adequate.

A group of 50 workers concerned with human cytogenetics met in 1971 on the occasion of the Fourth International Congress of Human Genetics in Paris to agree upon a uniform system of human chromosome identification. Their objective was accomplished and extended by the appointment of a standing committee, chaired by Dr. John Hamerton, which met initially in Edinburgh in January, 1972, and then with a number of expert consultants at Lake Placid in New York in December, 1974, and again in Edinburgh in April, 1975.

The 1971 meeting in Paris, together with the 1972 Edinburgh meeting of the standing committee, resulted in the report of the Paris Conference (1971), a highly significant document in the annals of human cytogenetics. This document proposed the basic system for designating not only individual chromosomes but also chromosome regions and bands, and it provided a way in which structural rearrangements and variants could be described in terms of their band composition.
By 1974 it had become clear that the number of workers in the field was now too great to allow the holding of such conferences as the Chicago and Paris ones, where the majority of laboratories involved could be represented. The standing committee therefore proposed holding smaller, nonrepresentative conferences, each on a number of fairly specific topics and that would utilize expert consultants for each topic. The first meeting of this type was held in 1974 in Lake Placid and the second in 1975 in Edinburgh, at which a number of specific topics—including heteromorphic chromosomes of the Hominoidea, and chromosome registers—were discussed. These discussions were reported in the 1975 supplement to the Paris Conference report (Paris Conference [1971], Supplement [1975]).

A further change came about in 1976 at the Fifth International Conference of Human Genetics in Mexico City, when a meeting of all interested human cytogeneticists was held to elect an international standing committee on human chromosome nomenclature. These elections provided a truly international and geographic representation for the standing committee and provided a mandate to the committee to continue its work in proposing ways in which human chromosome nomenclature might be improved. Dr. Jan Lindsten was appointed the chairman of this committee.

The committee met in Stockholm in 1977 and, following past practice, invited a number of expert consultants to meet with it. It was decided at this meeting to cease labeling reports geographically and to unify the various conference reports reviewed above into the present document, entitled “An International System for Human Cytogenetic Nomenclature (1978),” to be ISCN (1978) abbreviated “ISCN (1978).” The ISCN (1978) includes all major decisions of the Denver, London, Chicago, and Paris Conferences, without any major changes but edited for consistency and accuracy. It thus provides in one document a complete system of human cytogenetic nomenclature that has stood the test of time and that should be of value not only to those entering the field for the first time but also to experienced cytogeneticists.

To avoid confusion, no major changes have been made in the Paris Conference (1971) report, and the original Paris diagram has been provided as an appendix. A new diagram is provided, based on G and R banding. In this diagram the location of some bands have been corrected and one band, 3p27, has been omitted. Some bands that were not included in the Paris diagram but which can now be seen with improved techniques are included in the present diagram but designated as sub-bands.

In addition to material from previous reports, the ISCN (1978) includes an entirely new section on a suggested nomenclature for acquired aberrations. The nomenclature for human meiotic chromosomes is expanded to allow for the inclusion of chiasma count and location, and in the face of severe criticisms of the hominoid nomenclature proposed in the Paris supplement, a new phylogenetic nomenclature is provided alongside the conventional nomenclature given in 1975. New data on the hominoids obtained since 1975 have allowed the correction of some of the problem areas identified in the 1975 report. Finally, all the terms used in this report are defined either in the text or in the latest edition of Rieger et al. (1976).

One major area of concern that has not formed the topic for a standing committee meeting to date, although a working party under the chairmanship of Dr. Bernard Dutrillaux was established in 1977, is a nomenclature system for the large number of bands that can be seen in artificially induced prophase-like chromosomes. This will, it is hoped, be the subject for a future meeting in
1979, by which time sufficient data should be available for specific recommendations to be made.

It is to be hoped that the compilation of a comprehensive nomenclature based on the last 22 years of experience with human cytogenetics will be of value and will greatly improve communication in the field. In conclusion, I should like to express the appreciation of those involved in this work to The National Foundation—March of Dimes for their consistent and substantial financial support over the past 12 years, and in particular to Dan Bergsma and the late Virginia Apgar for their encouragement and help, without which none of these developments would have been possible.

John L. Hamerton
June, 1978

Publications cited


ISCN (1978)

2 Nomenclature for Normal Chromosomes and Constitutional Chromosome Aberrations

2.1 INTRODUCTION

Human chromosome nomenclature is based on the results of several international conferences (Denver, 1960; London, 1963; Chicago, 1966; Paris, 1971; Paris [Supplement], 1975; Stockholm, 1977). This report, which results from the Stockholm Conference, summarizes the nomenclature as it has developed over the past 18 years into one source to facilitate communication of cytogenetic information and to eliminate former inconsistencies. In addition,
some new terms have been added in areas where warranted. Thus, this report supercedes previous nomenclature reports.

2.2 CHROMOSOME NUMBER AND MORPHOLOGY

2.2.1 Non-Banding Techniques

In the construction of the karyotype the autosomes are numbered from 1 to 22, as nearly as possible in descending order of length. The sex chromosomes are referred to as X and Y. The symbols p and q are used to designate, respectively, the short and long arms of each chromosome (see Table 3).

When the chromosomes are stained by methods which do not produce bands, they can be arranged into seven readily distinguishable groups based on descending order of size and the position of the centromere.

In contemporary publications the terms karyotype and idiogram have often been used indiscriminately. We would recommend that the term karyotype should be applied to a systematized array of the chromosomes of a single cell prepared either by drawing or by photography, with the extension in meaning that the chromosomes of a single cell can typify the chromosomes of an individual or even a species. The term idiogram would then be reserved for the diagrammatic representation of a karyotype, which may be based on measurements of the chromosomes.

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Group 1-3 (A) Large metacentric chromosomes readily distinguished from each other by size and centromere position

Group 4-5 (B) Large submetacentric chromosomes which are difficult to distinguish from each other

Group 6-12-X (C) Medium-sized metacentric chromosomes. The X chromosome resembles the longer chromosomes in this group. This large group is the one which presents major difficulty in identification of individual chromosomes without the use of banding techniques.

Group 13-15 (D) Medium-sized acrocentric chromosomes with satellites

Group 16-18 (E) Relatively short metacentric chromosomes (No. 16) or submetacentric chromosomes (Nos. 17 and 18)

Group 19-20 (F) Short metacentric chromosomes

Group 21-22-Y (G) Short acrocentric chromosomes with satellites. The Y chromosome is similar to these chromosomes but bears no satellites.

The group letter designations placed after the numbers are those agreed upon at the London Conference (1963). Not all chromosomes in the D and G groups need show satellites or secondary constrictions on their short arms in a single cell. The number and size of these structures are variable.

The following parameters are used to describe non-banded chromosomes: (1) the length of each chromosome, expressed as a percent of the total length of a normal haploid set, i.e., the sum of the lengths of the 22 autosomes and of the X chromosome; (2) the arm ratio of the chromosomes, expressed as the length of the longer arm relative to the shorter one; and (3) the centromeric index, expressed as the ratio of the length of the shorter arm to the whole length of the chromosome. The latter two indices are, of course, related algebraically. These measurements are presented in Appendix 1.

2.2.2 Chromosome Banding Techniques
2.2.2.1 Methods and Terminology
Numerous different technical procedures have been reported that produce banding patterns on metaphase chromosomes.
A band is defined as that part of a chromosome which is clearly distinguishable from its adjacent segments by appearing darker or lighter with one or more banding techniques. Bands that stain darkly with one method may stain lightly with other methods. The chromosomes are visualized as consisting of a continuous series of light and dark bands, so that, by definition, there are no “interbands.”
The methods first published for demonstrating bands along the chromosomes were those that used quinacrine mustard or quin-acrine dihydrochloride to produce a fluorescent banding pattern. These methods are named Q-staining methods and the resulting bands, Q-bands (Fig. 1). Other techniques which demonstrate identical bands along the chromosomes use the Giemsa dye mixture as the staining agent. These techniques are generally termed G-staining methods and the resulting bands, G-bands (Figs. 2 and 5). Some of the Giemsa techniques, however, give patterns that are opposite in staining intensity to those obtained by the G-staining methods, viz., the reverse-staining Giemsa methods, and the resulting bands are called R-bands (Fig. 5). Other fluorescent techniques were later developed that produce bands equivalent to both Q- and R-bands (Fig. 3).
The banding techniques fall into two principal groups: (1) those resulting in bands distributed along the length of the whole chromosome, such as G-, Q-, and R-bands, including techniques that demonstrate patterns of DNA synthesis, and (2) those that stain a restricted number of specific bands or structures. These include methods which reveal centromeric (constitutive heterochromatin) bands (C-bands) (Fig. 4), telomeric bands (T-bands), and nucleolus organizing regions (NORs) (Table 1).
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Fig. 1. The human karyotype: Q-banded with quinacrine dihydrochloride. (Courtesy of Drs. H.S. Wang, V. Niewczas, and J.L. Hamerton.)
The patterns obtained with the various C-banding methods, such as staining in Giemsa solution buffered at pH 11 (the G-H technique), do not permit identification of every chromosome in the somatic cell complement but, as demonstrated in Table 1, can be used to identify specific chromosomes. The C-bands on chromosomes 1, 9, and 16 and the large distal region of Yq are all associated with obvious morphological variability. The short-arm regions of the acrocentric chromosomes also demonstrate varia-

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322 (10)

Y

19 20 21 22
Fig. 2. The human karyotype: G-banded by treating with trypsin-EDTA and staining with Giemsa. The metaphase was selected to demonstrate bands seen in preparations of good quality and not to show all the bands which can be seen in more extended chromosomes. (Method of H.P. Klinger, 1972, courtesy of the author.)

2.2.2.2 X- and Y-Chromatin

Inactive X chromosomes, as well as Y chromosomes, appear as distinctive structures in interphase nuclei, for which the terms X-chromatin (Barr body, sex chromatin, X-body) and Y-chromatin (Y-body), respectively, should be used.

Fig. 3. The human karyotype: R-banded with acridine orange after heat treatment. (Courtesy of Drs. H.S. Wang, V. Niewczas, and J.L. Hamerton.)

2.2.2.3 Characterization of Chromosomes by Fluorescent Banding Techniques

The initial classification of human chromosomes is based on the length and centromere index used for non-banded chromosomes. The numbers assigned to each chromosome are based on the Q-banding pattern as given by Caspersson et al. (1971). The designation of the additional chromosome associated with

\[ \text{SCN (1978)} \]

\[
\begin{array}{cccccccccccccc}
\end{array}
\]

\[
\begin{array}{cccccccccccccc}
& & & \text{i} & & & & & & & & & \\
\text{19} & & & & & & & & & & & & & \\
\text{20} & & & & & & & & & & & & & \\
\text{21}
\end{array}
\]
The human karyotype: C-banded by treating with barium hydroxide, followed by SCC, and then stained with Giemsa. In this metaphase the chromosomes show faint G-bands in addition to C-bands, allowing identification of individual chromosomes. (Courtesy of Dr. P. Pearson and J. Garver.)

Down syndrome has been retained as No. 21, although it is now known to be smaller than No. 22.

Some mitoses show considerable nonuniformity, in that homologous chromosomes may differ greatly in overall fluorescent intensity and relative length. Identification must therefore be based on the fluorescent banding pattern of the individual chromosome rather than on its overall intensity. However, intensity may serve

### Table 1

| Chromosome number | Technique | 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9 | 10 | 11 | 12 | 13 | 14 | 15 | 16 | 17 | 18 | 19 | 20 | 21 | 22 | X |
|-------------------|-----------|---|---|---|---|---|---|---|---|---|---|----|----|----|----|----|----|----|----|----|----|----|----|----|----|
|                   |           |   |   |   |   |   |   |   |   |   |   |    |    |    |    |    |    |    |    |    |    |    |    |    |    |

Only the brilliant and variable Q-bands have been considered.
variable, \( p = \) short arm, \( q = \) long arm.

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as a secondary criterion, if due allowance is made for non-uniformity. The following terms are used to indicate the approximate intensity of fluorescence:

- **negative**: No or almost no fluorescence
- **pale**: As on distal lp
- **medium**: As the two broad bands on 9q
- **intense**: As the distal half of 13q
- **brilliant**: As on distal Yq

2.3 CHROMOSOME BAND NOMENCLATURE

2.3.1 Identification and Definition of Chromosome Landmarks, Bands, and Regions

Each chromosome in the human somatic cell complement is considered to consist of a continuous series of bands, with no unhand ed areas. As defined earlier, a band is a part of a chromosome clearly distinguishable from adjacent parts by virtue of its lighter or darker staining intensity. The bands are allocated to various regions along the chromosome arms, and the regions are delimited by specific landmarks. These are defined as consistent and distinct morphologic features important in identifying chromosomes. Landmarks include the ends of the chromosome arms, the centromere, and certain bands. The bands and the regions are numbered from the centromere outward. A region is defined as any area of a chromosome lying between two adjacent landmarks.

2.3.2 Designation of Regions and Bands

Regions and bands are numbered consecutively from the centromere outward along each chromosome arm. Thus, the two regions adjacent to the centromere are labeled as 1 in each arm; the next, more distal regions as 2, and so on. A band used as a landmark is considered as belonging entirely to the region distal to the landmark and is accorded the band number of 1 in that region.

In designating a particular band, four items are required:

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1. the chromosome number, 2. the arm symbol, 3. the region number, and 4. the band number within that region. These items are given in order without spacing or punctuation. For example, lp33 indicates chromosome 1, short arm, region 3, band 3.

Diagrammatic Representation of Landmarks and Bands

The original banding pattern was described in the Paris Conference (1971) report and was based on the patterns observed in different cells stained with either the Q-, G-, or R-band technique (Appendix 2). The banding patterns obtained with these staining methods agreed sufficiently to allow the construction of a single diagram representative of all three techniques, although the position of heterochromatin adjacent to the centromere was indicated on the basis of the Q-band technique only. The diagram was not based on measurements, nor on sequentially banded cells. The length and position of the chromosome bands and the relative band sizes and distributions can, however, be taken to be approximately correct. The bands were designated on the basis of their midpoints and not by their margins. No attempt was made to indicate the intensity of fluorescence or staining, because this will vary with different techniques. Intensity was taken into consideration, however, in determining which bands should serve as landmarks on each chromosome in order to divide the chromosome into natural, easily recognizable morphologic
regions. A list of bands serving as landmarks which were used in constructing this diagram is provided in Table 2. A new representation of chromosome bands based on G- and R- staining methods, which takes staining intensity into account, and in which the bands are numbered according to the Paris Conference (1971) nomenclature, is presented in Fig. 5, as well as in the larger fold-out attached to the back cover.

Subdivision of an Existing Landmark or Band
In the event that a band serving as a landmark requires subdivision, all sub-bands derived from it retain the original region.

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Fig. 5. Photographs of G- and R-banded human metaphase chromosomes and their diagrammatic representations. The chromosomes were selected to demonstrate bands seen in preparations of good quality and not to show all the bands which can be seen in more extended chromosomes. The relative position of some bands differs from those shown in the diagram of the Paris Conference (1971) report, reproduced here as Appendix 2. Bands 8q21, 19ql3, XpII, and Xp22 have been subdivided, and band 3p27 has been omitted. (Prepared by Drs. B. Dutrillaux and M. Prieur. G-banding according to the
method of Couturier and Lejeune, 1976; R-banding according to Dutrillaux et al., 1975.) (A larger version of this figure appears as a fold-out attached to the back cover.)

Table 2 Bands serving as landmarks which divide the chromosomes into cytologically defined regions. The omission of an entire chromosome or chromosome arm indicates that either both arms or the arm in question consists of only one region, delimited by the centromere and the end of the chromosome arm.

<table>
<thead>
<tr>
<th>Chromosome</th>
<th>Number</th>
<th>Landmarks</th>
</tr>
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<tbody>
<tr>
<td>2</td>
<td>P</td>
<td>a 2</td>
</tr>
<tr>
<td>3</td>
<td>P</td>
<td>a 2</td>
</tr>
<tr>
<td>4</td>
<td>a</td>
<td>3</td>
</tr>
<tr>
<td>5</td>
<td>q</td>
<td>3</td>
</tr>
<tr>
<td>6</td>
<td>P</td>
<td>2</td>
</tr>
<tr>
<td>7</td>
<td>P</td>
<td>a 2</td>
</tr>
<tr>
<td>8</td>
<td>P</td>
<td>q 3</td>
</tr>
<tr>
<td>9</td>
<td>a</td>
<td>2</td>
</tr>
<tr>
<td>10</td>
<td>q</td>
<td>2</td>
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<tr>
<td>11</td>
<td>a</td>
<td>2</td>
</tr>
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<td>12</td>
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<td>2</td>
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<td>13</td>
<td>a</td>
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<td>14</td>
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<td>18</td>
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<td>2</td>
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<tr>
<td>21</td>
<td>a</td>
<td>2</td>
</tr>
<tr>
<td>X</td>
<td>P</td>
<td>a 2</td>
</tr>
</tbody>
</table>

Proximal band of medium intensity (21), median band of medium intensity (31)
Proximal negative band (21) distal to variable region, median intense band (31), distal medium band (41)
Median negative band (21)
Proximal negative band (21), distal negative band (31)
Median negative band (21)
Proximal negative band (21), distal negative band (31)
Median band of medium intensity (21), distal negative band (31)
Median negative band (21)
Median negative band (21)
Distal medium band (21)
Proximal medium band (21), median band of medium intensity (31)
Median negative band (21)
Median band of medium intensity (21)
Median intense band (21)
Median band of medium intensity (21), distal band of medium intensity (31)
Proximal intense band (21)
Median negative band (21)
Median band of medium intensity (21)
Median intense band (21)
Median band of medium intensity (21), distal intense band (31)
Proximal intense band (21), distal medium band (31)
Median intense band (21)
Median band of medium intensity (21)
Proximal negative band (21)
Median negative band (21)
Median intense band (21)
Proximal medium band (21)
Proximal medium band (21)
Normal Chromosomes and Constitutional Aberrations 331 (19)
REGION BAND
(a)
4 1

D3.3 | 3.3
■ ■ ■

2 2

D

Fig. 6b. Method for numbering the subdivisions of a landmark bridging two regions: (A) the original landmark (band 31); (B) the subdivision of band 31 into three equal bands 31.1, 31.2, and 31.3; (C) alternatively, the subdivision of band 31 into three unequal bands; (D) further subdivision of band 31.3 into three equal bands 33.31, 33.32, and 33.33.

(b) 332 (20)
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and band number of that landmark (Fig. 6a). This rule is followed even if subdivision causes one or more sub-bands to lie in an adjacent region.

Whenever an existing band is subdivided, a decimal point is placed after the original band designation and is followed by the number assigned to each sub-band. The sub-bands are numbered sequentially from the centromere outward. For example, if the original band lp33 is subdivided into three equal or unequal sub-bands, the sub-bands are labeled lp33.1, lp33.2, and lp33.3, sub-band 33.1 being proximal and 33.3 distal to the centromere (Fig. 6b). If a sub-band is subdivided, additional digits, but no further punctuation, are used; e.g., sub-band lp33.1 might be further subdivided into lp33.11, lp33.12, etc. (Fig. 6). See also the method proposed for specification of a break point within a band in Section 2.4.4.2.

2.4 NOMENCLATURE SYMBOLS

A system of nomenclature symbols is proposed for describing the human chromosomes and their aberrations. A list of these symbols is given in Table 3.

In the description of a karyotype the first item to be recorded is the total number of chromosomes, including the sex chromosomes, followed by a comma (,). The sex chromosome constitution is given next.

2.4.1 Normal Karyotypes

Normal human karyotypes are designated as follows:

46, XX Normal female 46, XY Normal male

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Table 3

List of symbols and abbreviated terms. The abbreviations may be used in any combination considered useful. Section references are given within parentheses for those terms which are defined in greater detail in the text. Other terms are defined in Rieger et al. (1976).

<table>
<thead>
<tr>
<th>Symbol</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>A1</td>
<td>First meiotic anaphase</td>
</tr>
<tr>
<td>AII</td>
<td>Second meiotic anaphase</td>
</tr>
<tr>
<td>ace</td>
<td>Acentric fragment (see also /)</td>
</tr>
<tr>
<td>arson</td>
<td>From (§ 2.4.4.4)</td>
</tr>
<tr>
<td>asterisk (*)</td>
<td>Used like a multiplication sign (§ 6)</td>
</tr>
<tr>
<td>b</td>
<td>Break</td>
</tr>
<tr>
<td>cen</td>
<td>Centromere</td>
</tr>
<tr>
<td>chi</td>
<td>Chimera</td>
</tr>
<tr>
<td>col singled (:)</td>
<td>Break in detailed descriptions, §</td>
</tr>
<tr>
<td>col double (::)</td>
<td>Break and reunion (in detailed descriptions, §2444)</td>
</tr>
<tr>
<td>cs</td>
<td>Chromosome</td>
</tr>
<tr>
<td>ct</td>
<td>Chromatid</td>
</tr>
<tr>
<td>ex</td>
<td>Complex</td>
</tr>
<tr>
<td>del</td>
<td>Deletion</td>
</tr>
<tr>
<td>der</td>
<td>Derivative chromosome (§ 2.4.7)</td>
</tr>
<tr>
<td>dia</td>
<td>Diakinesis</td>
</tr>
<tr>
<td>die</td>
<td>Dicentric</td>
</tr>
<tr>
<td>dip</td>
<td>Diploptene</td>
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<td>dir</td>
<td>Direct</td>
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<td>dis</td>
<td>Distal</td>
</tr>
<tr>
<td>dit</td>
<td>Dictate</td>
</tr>
<tr>
<td>dmi</td>
<td>Double minute (§ 4.2.1)</td>
</tr>
<tr>
<td>dup</td>
<td>Duplication</td>
</tr>
<tr>
<td>e</td>
<td>Exchange</td>
</tr>
<tr>
<td>end</td>
<td>Endoreduplication</td>
</tr>
<tr>
<td>equ sign ( = )</td>
<td>Sum of (§ 3.2.2 and § 5.1)</td>
</tr>
<tr>
<td>Anastomoses</td>
<td>Description</td>
</tr>
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<td>-------------</td>
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</tr>
<tr>
<td>Fragment (see also ace)</td>
<td>Female</td>
</tr>
<tr>
<td>Gap</td>
<td>Secondary constriction Isochromosome</td>
</tr>
<tr>
<td>Insertion</td>
<td>Inversion</td>
</tr>
<tr>
<td>Leptotene</td>
<td>First meiotic metaphase</td>
</tr>
<tr>
<td>Male</td>
<td>Second meiotic metaphase</td>
</tr>
<tr>
<td>Marker chromosome</td>
<td>Male</td>
</tr>
<tr>
<td>Maternal origin</td>
<td>Median</td>
</tr>
<tr>
<td>Minute</td>
<td>Loss of (§ 2.4.2.1)</td>
</tr>
<tr>
<td>Modal number</td>
<td>Mosaic</td>
</tr>
<tr>
<td>Oogonial metaphase</td>
<td>Short arm of chromosome</td>
</tr>
<tr>
<td>First meiotic prophase</td>
<td>Pachytene</td>
</tr>
<tr>
<td>Pseudoviviparous</td>
<td>Chromosome or chromosome structure</td>
</tr>
<tr>
<td>Oligosomatid</td>
<td>Paternal origin</td>
</tr>
<tr>
<td>Premature chromosome condensation</td>
<td>Philadelphia chromosome</td>
</tr>
<tr>
<td>Gain of (§ 2.4.2.1)</td>
<td>Proximal</td>
</tr>
<tr>
<td>Pseudo</td>
<td>Quadriradial</td>
</tr>
<tr>
<td>Pulverization</td>
<td>Indicates questionable identification</td>
</tr>
<tr>
<td>Long arm of chromosome</td>
<td>Chromosome or chromosome structure</td>
</tr>
<tr>
<td>Quadriradial</td>
<td>Paternal origin</td>
</tr>
<tr>
<td>Ring chromosome</td>
<td>Maternal origin</td>
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<td>Median</td>
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<td>Recombinant chromosome</td>
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<td>Robertsonian translocation</td>
<td>Modal number</td>
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<td>Mosaic</td>
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<td>Sister chromatid exchange</td>
<td>Oogonial metaphase</td>
</tr>
<tr>
<td>Side-line. sub-line</td>
<td>Short arm of chromosome</td>
</tr>
<tr>
<td>Separates chromosomes and regions in structural rearrangements involving more than one chromosome (§ 2.4.3 and § 2.4.4.1)</td>
<td>Proximal</td>
</tr>
<tr>
<td>Stem-line</td>
<td>Chromosome or chromosome structure</td>
</tr>
<tr>
<td>Separates cell lines in describing or chimeras (§ 2.4.4.2)</td>
<td>Long arm of chromosome</td>
</tr>
<tr>
<td>Spermatogonial metaphase</td>
<td>Quadriradial</td>
</tr>
<tr>
<td>Translocation</td>
<td>Maternal origin</td>
</tr>
<tr>
<td>Tandem translocation</td>
<td>Paternal origin</td>
</tr>
</tbody>
</table>
Numerical Chromosome Aberrations

Numerical chromosome aberrations can be described as follows:

- **45, X** 45 chromosomes, one X chromosome
- **47, XXY** 47 chromosomes, XXY sex chromosomes
- **49, XXXXY** 49 chromosomes, XXXXY sex chromosomes

**Use of + and – Signs**

The + or – signs are placed before the appropriate symbol to indicate additional or missing whole chromosomes. They are placed after a symbol to indicate an increase or decrease in the length of a chromosome, an arm, or a region of a chromosome. The autosomes are specified only when an abnormality is present. Thus, if there is a numerical aberration of the autosomes, the group letter or number of the extra or missing autosome, preceded by a ÷ or – sign, follows the sex chromosome designation, e.g.:

- **45, XX, —C** 45 chromosomes, XX sex chromosomes, a missing C-group chromosome
- **48, XXY, + G** 48 chromosomes, XXY sex chromosomes, an additional G-group chromosome
- **47, XY, + 21** 47 chromosomes, XY sex chromosomes, an additional chromosome 21
- **46, XY, +18, -21** 46 chromosomes, XY sex chromosomes, an extra chromosome 18 and a missing chromosome 21

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- **46, XY, lq+** Male karyotype with 46 chromosomes showing an increase in the length of the long arm of one chromosome 1
- **47, XY, +14p+** Male karyotype with 47 chromosomes, including an additional chromosome 14 which has an increase in the length of its short arm

A question mark (?) is used to indicate uncertainty. If it is suspected that a missing or extra chromosome belongs to a particular group, but this is not certain, the question mark may precede the group designation or, in some cases, the chromosome number, e.g.:

- **45, XX, —?8** 45 chromosomes, XX sex chromosomes, a missing chromosome which is probably chromosome 8

As another example, the description of the karyotype of an X-chromatin positive female with an additional small acrocentric chromosome could be written, depending on the amount of available information, **47, XX, + ?G, 47, XX, + G, 47, XX, + ?21, or 47, XX, +21.**

A triploid or polyploid cell should be evident from the chromosome number and from the further designations, e.g., 69, XXY; 70, XXY, + G. An endoreduplicated metaphase is indicated by preceding the karyotype designation with the abbreviation end, e.g., end46, XX. If multiplicity of
endoreduplications are to be indicated, an Arabic numeral is used before end to indicate this, e.g.,
2end46, XX; 4end46, XX.

2.4.2.2 Chromosome Mosaics and Chimeras
The chromosome constitution of the different cell lines in a mosaic or chimera are listed in
numerical order, irrespective of the frequencies of the cell types in the individual studied. The
karyotype designations are separated by a slant line, or solidus (/) (see also Section 4.5), e.g.:

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45, X/46, XY  A chromosome mosaic with two cell lines,
one with 45 chromosomes and a single X, the other with 46 chromosomes and XY sex
chromosomes

46, XY/47, XXY, +G  A chromosome mosaic with a normal male
cell line and a cell line with an extra G-group chromosome

45, X/46, XX/47, XXX  A triple cell line mosaic

In order to distinguish between chromosome mosaics (cell lines originating from the same
zygote) and chimeras (cell lines originating from different zygotes), it is recommended that the
triplets mos and chi be used, as follows:

mos45, X/46, XY  A chromosome mosaic with two cell lines,
one with 45 chromosomes and a single X, the other with 46 chromosomes and XY sex
chromosomes

chi46, XX/46, XY  A chimera with both XX and XY cell
lines

In most instances the triplets mos and chi will be needed only for the initial description in any
report; subsequently, the simple karyotype designation may be used (see also Section 4.5).

Structural Chromosome Aberrations in Non-Banded Chromosomes
All symbols for structural aberrations are to be placed before the designation of the chromosome
or chromosomes involved. The rearranged chromosome or chromosomes should always be
placed in parentheses.

Pericentric inversions: The result of a pericentric inversion is indicated by $p^+q^-$ or $p^-q^+$,
which is enclosed in parentheses and preceded by the abbreviation inv, e.g., inv(Dp + q —).

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Translocations: A translocation is indicated by the letter t followed by parentheses which include
the chromosomes involved, e.g.:

46, XY, t(Bp—;Dq +) or A balanced reciprocal translocation
46, XY, t(Bp+,Dq —) between the short arm of a B- and
the long arm of a D-group chromosome

Translocations involving a sex chromosome and an autosome would be designated as:

46, X, t(Xq+;16p —) A reciprocal translocation between
the long arm of an X chromosome and the short arm of a chromosome 16 in a female
46, Y, t(Xq + ;16p —) The same translocation in a male
46, X, t(Yp+;16p —) A reciprocal translocation between
the short arm of a Y chromosome and the short arm of a chromosome 16

The remaining normal sex chromosome is written in its usual position after the chromosome
number, and the other sex chromosome which is involved in the translocations is included in
parentheses preceding the autosome concerned.
The separation of the chromosomes within the parentheses by a semicolon (;) indicates that two structurally altered chromosomes are present and that the translocation is balanced. In a centric fusion type of translocation, in which only one translocation chromosome is present, the semicolon is omitted, e.g.:

45, XX,— D, — G, + t(DqGq) 45 chromosomes, XX sex chromosomes, one chromosome missing from the D group and one from the G group, their long arms having united to form a translocation chromosome.

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If desired, karyotype descriptions of balanced Robertsonian translocations, as well as whole-arm translocations, may be shortened, e.g., 45, XX, t(DqGq). Unbalanced karyotypes, however, should be written out completely:

46, XX, — D, + t(DqGq) 46 chromosomes, XX sex chromosomes, one chromosome missing from the D group. The long arm of the D chromosome is united with the long arm of a G chromosome. Since there are four normal G chromosomes, part of a G is present in triplicate.

If, as rarely happens, a small chromosome is present as well, implying a reciprocal translocation, the karyotype could be written 46, XX, — D, — G, + t(DpGp), + t(DqGq).

When it is known that a particular chromosome has been inherited from the mother or the father, this may be indicated by the abbreviation mat or pat. For instance, if a father is carrying a balanced reciprocal translocation, 46, XY, t(Bp —; Dq +), and his malformed son has inherited one of the two abnormal chromosomes, the son’s complement is written as 46, XY, Bp — pat or 46, XY, Dq+pat, depending on which abnormal chromosome has been transmitted. If the son has inherited both chromosomes involved in the translocation, his complement would be expressed as 46, XY, t(Bp —; Dq +) pat.

A ring chromosome is designated as:

46, XX, r(16) Female karyotype with 46 chromosomes, including a ring chromosome 16

Isochromosomes or dicentrics are designated as:

46, X, i(Xq) Female karyotype with 46 chromosomes, including one normal X chromosome and one chromosome represented by an isochromosome for the long arm of the X

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46, X, dic(Y) Karyotype with 46 chromosomes, one X chromosome, and a dicentric Y chromosome

In the interests of clarity, complex rearrangements necessitating lengthy descriptions should be written out in full the first time they are used in a report. An abbreviated version may be used subsequently, providing it is clearly defined immediately after the complete notation.

2.4.4 Designating Structural Chromosome Aberrations by Breakage Points and Band Composition

Two systems for designating structural abnormalities are presented. One is a short system in which the nature of the rearrangement and the break point or points are identified by the bands or regions in which the breaks occur. Because of the conventions built into this system, the band composition of the abnormal chromosomes present can be readily inferred from the information provided in the symbolic description. The other is a detailed system which, besides identifying the type of rearrangement, defines each abnormal chromosome present in terms of its band
composition. The two systems are not mutually exclusive. The notation used to identify the rearrangement and the method of specifying the break points are common to both systems and will be presented first.

2.4.4.1 Specification of Chromosome Rearrangements

Single and three-letter designations are used to specify rearranged (i.e., structurally altered) chromosomes. The number of the chromosome involved in the change is specified within parentheses immediately following the symbol identifying the type of rearrangement, e.g., r(18); inv(2). If two or more chromosomes have been altered, a semicolon (;) is used to separate their designations. If one of the rearranged chromosomes is a sex chromosome, then it is listed first; otherwise, the chromosome having the lowest chromosome number is always specified first, e.g., t(X;3); t(2;5).

The only exception to this rule involves certain three-break rearrangements in which part of one chromosome is inserted at a point of breakage in another chromosome. In this event, the receptor chromosome is specified first, regardless of whether it is a sex chromosome or an autosome with a number higher or lower than that of the donor chromosome.

For translocations involving three separate chromosomes, with one break point in each chromosome, the rule is still followed that the sex chromosome or autosome with the lowest number is specified first. The chromosome listed next is the one that receives a segment from the first chromosome, and the chromosome specified last is the one that donates a segment to the first listed chromosome (see also “Complex Translocations,” Section 2.4.4.4).

Designations required to specify rearrangements are listed in Table 3 and explained below.

Deletions: The abbreviation del is used to designate a chromosome deletion.

Translocations: All translocations are specified by the symbol t. If the type of translocation, i.e., Robertsonian, reciprocal, or tandem, is to be emphasized, t may be replaced with rob, rep, or tan, respectively. The symbol rep is used for reciprocal translocations to avoid confusion with rec, which is used to designate a recombinant chromosome. Translocations resulting in a dicentric chromosome are designated simply by die.

Three-break rearrangements: These may involve one, two, or three chromosomes. Rearrangements involving three or more chromosomes will be referred to as complex translocations. Several terms have been employed in the cytogenetic literature for three-break rearrangements involving one or two chromosomes; these include shift, insertion, and transposition. In this report, all three-break rearrangements involving one or two chromosomes are referred to as insertions since they result from the excision of a segment following two breaks in one chromosome arm and its insertion at a point of breakage in either the same arm, the opposite arm of the same chromosome, or in another chromosome. The order of the bands on the inserted segment in relation to the centromere at the new site may be the same as at the original site (direct insertion) or may be reversed (inverted insertion). The abbreviation dir ins is used to indicate a direct insertion and inv ins to indicate an inverted insertion.

2.4.4.2 Specification of Break Points

The location of any given break is specified by the band in which that break has occurred. Since it is not possible at present to define band interfaces accurately, a break suspected at an interface
between two bands is identified arbitrarily by the higher of the two band numbers, i.e., the number of the band more distal to the centromere.

A given break may sometimes appear to be located in either of two consecutive bands. A similar situation may occur when breaks at or near an interface between two bands are studied with two or more techniques. In this event, the break can be specified by both band numbers separated by the word or, e.g., lq23 or 24, indicating a break in either band lq23 or band lq24. If a break can be localized to a region but not to a particular band, only the region number should be specified, e.g., lp1, instead of lp12 or 12 or 13. If the break point can be assigned only to two adjacent regions, both suspected regions should be specified, e.g., 1q2 or 3.

When a break point can be specified within a band, a numbering system can be used to designate the location of break points on the basis of the relative distance of the break point from the proximal margin of the band concerned. The interface between two bands is arbitrarily designated by the higher of the two band numbers followed by 00, as follows:
lpl200 The interface between the bands lpll and lpl2
lp1206 A point six-tenths of the distance from the proximal edge of band lpl2

Note that this allows the division of a band into 10 units. Two digits should always be used, the first of which will always be zero (0), which distinguishes the designation of a break point from the method for subdividing bands (see Section 2.3.4).

2.4.4.3 Short System for Designating Structural Chromosome Aberrations

In this system, structurally altered chromosomes are defined only by their break points. The break points are specified within parentheses immediately following the designation of the type of

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rearrangement and the chromosome(s) involved, as described earlier. The break points are identified by band designations. For example, del(l)(q21) defines a terminal deletion in the long arm of chromosome 1 resulting from a break at band lq21.

Two-break rearrangements: When both arms of a single chromosome are involved in a two-break rearrangement, the break point in the short arm is always specified before the break point in the long arm; e.g., inv(2)(p21q31) defines a pericentric inversion in chromosome 2 with break points in bands 2p21 and 2q31. When the two breaks occur within the same arm, the break point more proximal to the centromere is specified first; e.g., inv(2)(pl3p23) defines a paracentric inversion in the short arm of chromosome 2 with break points in bands 2pl3 and 2p23.

Three-break rearrangements: When an insertion within a single chromosome occurs, the break point at which the chromosome segment is inserted is always specified first. The remaining break points are specified in the same way as in a two-break rearrangement; i.e., the more proximal break point of the inserted segment is specified first and the more distal one last.

“Proximal” and “distal” refer here to the positions of the break points following the rearrangement and not to their original positions. For example, inv ins(2)(ql3p23pl3) defines an inverted insertion in chromosome 2 of the short-arm segment lying between bands 2pl3 and 2p23 into the long arm at band 2ql3. Because the insertion is inverted, band 2p23 is now proximal and band 2pl3 distal to the centromere.

Rearrangements affecting two or more chromosomes: The break points are specified in the same order as the chromosomes involved are specified, and a semicolon is used to separate the break points (punctuation is never used to separate break points in the same chromosome). For
example, rcp(2;5)(q21;q31) defines a reciprocal translocation between the long arms of chromosomes 2 and 5, with break points at bands 2q21 and 5q31.

Detailed System for Designating Structural Chromosome Aberrations

Structurally altered chromosomes are defined by their band composition. The conventions used in the short system are

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retained in the detailed system, except that an abbreviated description of the band composition of the rearranged chromosome or chromosomes is specified within the final pair of parentheses, instead of only the break points.

Additional symbols: A single colon (:) is used to indicate a chromosome break and a double colon (::) to indicate break and reunion. In order to avoid an unwieldy description, an arrow (→), meaning from—to, is employed. The end of a chromosome arm may be designated either by its band designation or by the symbol ter, meaning terminal, preceded by the arm designation; e.g., pter indicates the end of the short arm and qter, the end of the long arm. When it is necessary to indicate the centromere, the abbreviation cen should be used.

Designating the band composition of a chromosome: The description starts at the end of the short arm and proceeds through to the end of the long arm, with the bands being identified in the order in which they occur in the rearranged chromosome. If the rearrangement is confined to a single chromosome, the chromosome number is not repeated in the band description. If more than one chromosome is involved, however, the bands and chromatid ends are identified with the appropriate chromosome numbers.

If, owing to a rearrangement, no short-arm segment is present at the end of either arm, the description of the structurally rearranged chromosome starts at the end of the long-arm segment with the lowest chromosome number.

Where more than one chromosome is involved, the chromosome descriptions are presented in the same numerical order as the chromosomes involved in the rearrangement. In the special case of an unbalanced reciprocal translocation between the long arm of one chromosome and the short arm of another, the derivative chromosome carrying the centromere belonging to the chromosome with the lower chromosome number is described first. In the special case of complex translocations involving three or more chromosomes, note the departure from the rules given under “Complex Translocations” and in Section 4.2.4.

In all the examples presented in this section, the short system designation is shown first and the detailed system second, followed by a brief explanation of the latter.

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Isochromosomes
46, X,i(Xq)
46, X, i(X)(pter→cen→qter)

Break points in this type of rearrangement are at or close to the centromere and cannot be specified. The designation indicates that both entire long arms of the X chromosome are present and are separated by the centromere.

Terminal Deletions
46, XX, del(l)(q21)
46, XX, del(l)(pter→q21)
The single colon (:) indicates a break at band lq21 and deletion of the long-arm segment distal to it. The remaining chromosome consists of the entire short arm of chromosome 1 and part of the long arm lying between the centromere and band lq21.

**Interstitial Deletions**

46, XX, del(l)(q21q31)

The double colon (::) indicates breakage and reunion of bands lq21 and lq31 in the long arm of chromosome 1. The segment lying between these bands has been deleted.

**Paracentric Inversions**

46, XY, inv(2)(p13p24)

Breakage and reunion have occurred at bands 2p13 and 2p24 in the short arm of chromosome 2. The segment lying between these bands is still present but inverted, as indicated by the reverse order of the bands with respect to the centromere in this segment of the rearranged chromosome.

**Pericentric Inversions**

46, XY, inv(2)(p21q31)

Breakage and reunion have occurred at band 2p21 in the short arm and 2q31 in the long arm of chromosome 2. The segment lying between these bands is inverted.

**Duplication of a Chromosome Segment**

The symbol dup is used. It can be preceded by the triplets dir or inv to indicate if the duplication is direct or inverted, e.g.:

46, XX, inv dup(2)(p23→pl4)

Breakage and reunion have occurred at band 2p23 of chromosome 2, with the segment p23→pl4 being inverted and duplicated.

**Ring Chromosomes**

46, XY, r(2)(p21q31)

Breakage has occurred at band 2p21 in the short arm and 2q31 in the long arm of chromosome 2. With deletion of the segments distal to these bands, the broken ends have joined to form a ring chromosome. Note the omission of the colon or double colon.

**Dicentric Chromosomes**

46, X, dic(Y)(ql2)

Breakage and reunion have occurred at band Yq12 on sister chromatids to form a dicentric Y chromosome. The alternative form of a dicentric Y chromosome might be:

46, X, dic(Y)(p1l)

Reciprocal Translocations
Breakage and reunion have occurred at bands 2q21 and 5q31 in the long arms of chromosomes 2 and 5, respectively. The segments distal to these bands have been exchanged between the two chromosomes. The derivative chromosome with the lowest number (i.e., chromosome 2) is designated first.

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46, XY, t(2;5)(p12;q31)

46, XY, t(2;5)(qter→2p2::5q31→5qter; 5pter→5q3::2p2→2qter)

Breakage and reunion have occurred at band 2p2 in the short arm and band 5q31 in the long arm of chromosomes 2 and 5, respectively. The segments distal to these bands have been exchanged between the two chromosomes. Note that the derivative chromosome bearing the centromere of the original chromosome 2 has no terminal short-arm segment, and, therefore, its description starts with the long-arm end having the lowest number, i.e., 2qter.

Robertsonian Translocations

45, XX, t(13;14)(p12;q11)

45, XX, t(13;14)(qter→3p1::14q11→14qter)

Breakage and reunion have occurred at band 13p1 in the short arm and band 14q11 in the long arm of chromosomes 13 and 14, respectively. The segment distal to band 14q11 has been translocated onto chromosome 13 at band 13p1. The rest of chromosome 14, with its centromere, has been lost along with the original segment distal to band 13p1, i.e., 13pter→13p1.

45, XX, t(13q14q)

45, XX, t(13;14)(qter-→cen→14qter)

Breakage has occurred at or near the centromeres of chromosomes 13 and 14. The rearranged chromosome has the long arms of both chromosomes separated by a centromere, the origin of which might have been either chromosome. Both short arms have been lost.

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45, XX, dic(13;14)(p11;pll)

45, XX, dic(13;14)(qter→3p1::14q11→14qter)

Breakage and reunion have occurred at bands 13p1 and 14q11 in the short arms of chromosomes 13 and 14, respectively. The segments distal to these bands have been deleted, and the remaining segments have joined at the break points in the short arms to form a dicentric translocation chromosome.

Whole-arm Translocations

Whole-arm exchanges that involve nonacrocentric chromosomes in which the position of the break points relative to the centromeres is not known can be described, e.g., as:

46, XY, t(2;3)(2p3p;2q3q)

46, XY, t(2;3)(2qter→cen→3pter;2qter→cen→3qter)

In this example, the entire short arms of chromosomes 2 and 3 have been exchanged, as well as the entire long arms of these chromosomes. The derivative chromosomes would be der(2p3p) and der(2q3q). The alternative rearrangement would be:

46, XY, t(2;3)(2p3q;2q3p)

46, XY, t(2;3)(2pter→cen→3qter;2qter→cen→3pter)

The two derivative chromosomes would then be der(2p3q) and der(2q3p).

In both cases the derivative chromosomes are designated according to their arms, not their centromeres. If it becomes possible to designate the centromere specifically as being derived
from one or the other of the chromosomes involved, this could be indicated by the symbol cen before the chromosome number,

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e.g., cen2, if it is known that the centromere was derived from chromosome 2. For example:

46, XY, t(2;3)(2pter→cen2→3qter;2qter→cen3→3pter)

This indicates that the origin of the centromere in both derivative chromosomes is known.

**Direct Insertions Within a Chromosome**

46, XY, dir ins(2)(pl3q21q31)

46, XY, dir ins(2)(pl3q21q31)

Breakage and reunion have occurred at band 2p3 in the short arm and bands 2q21 and 2q31 in the long arm of chromosome 2. The long-arm segment between bands 2q21 and 2q31 has been inserted into the short arm at band 2p3. The original orientation of the inserted segment has been maintained in its new position; i.e., band 2q21 remains more proximal to the centromere than band 2q31.

**Inverted Insertions Within a Chromosome**

46, XY, inv ins(2)(pl3q31q21q31)

46, XY, inv ins(2)(pl3q31q21q31)

Breakage and reunion have occurred at the same bands as in the previous example, and the insertion is the same except that the inserted segment has been inverted; i.e., band 2q21 in the inserted segment is now more distal to the centromere than band 2q31. The orientation of the bands within the segment has thus been reversed with respect to the centromere.

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**Direct Insertions Between Two Chromosomes**

46, XY, dir ins(5;2)(pl4;q22q32)

46, XY, dir ins(5;2)(pl4;q22q32)

Breakage and reunion have occurred at band 5p4 in the short arm of chromosome 5 and bands 2q22 and 2q32 in the long arm of chromosome 2. The long-arm segment between bands 2q22 and 2q32 has been inserted into the short arm of chromosome 5 at band 5p4. The original orientation of the inserted segment has been maintained in its new position, i.e., 2q22 remains more proximal to the centromere than 2q32. Note that the recipient chromosome is specified first.

**Inverted Insertions Between Two Chromosomes**

46, XY, inv ins(5;2)(pl4;q22q32)

46, XY, inv ins(5;2)(pl4;q22q32)

Breakage and reunion have occurred at the same bands as in the previous example, and the insertion is the same except that the inserted segment has been inverted; i.e., band 2q22 is now more distal to the centromere of the recipient chromosome than 2q32.

**Complex Translocations**

46, XX, t(2;5;7)(p21;q23;q22)

46, XX, t(2;5;7)(p21;q23;q22)

Breakage and reunion have occurred at band 2p21 in the short arm of chromosome 2 and at bands 5q23 and 7q22 in the long arm of chromosome 7. This indicates that the origin of the centromere in both derivative chromosomes is known.
arms of chromosomes 5 and 7, respectively. The segment of chromosome 2 distal to 2p21 has
been translocated onto chromosome 5 at 5q23, the segment of chromosome 5 distal to 5q23 has
been translocated onto chromosome 7 at 7q22, and the segment of chromosome 7 distal to 7q22
has been translocated onto chromosome 2 at 2p21. Note that the chromosome specified first is
the one with the lowest number, the chromosome specified next is the one receiving a segment
from the first one listed, and the chromosome specified last is the one donating a segment to the
first chromosome listed.

Four-Break Rearrangements

There are a very large number of possible four-break rearrangements. These can be described
using the conventions outlined here. A single example from a known case is illustrated here to
indicate how such rearrangements can be handled—a double reciprocal translocation involving
three chromosomes (see also Section 4.2.4):

46, XX, t(l;3)(3;9)(pl2:p13q25;q22)
46, XX, t(l;3)(3;9)(lqter→lpl2::3pl3→3pter;
Ipter→lpl2::3pl3→3q25::9q22→9qter; 9pter→9q22::3q25→3pter)

Breakage and reunion have occurred at bands lpl2 and 3pl3 in the short arms of chromosomes 1
and 3, respectively, and at 3q25 and 9q22 in the long arms of chromosomes 3 and 9,
respectively. The segments distal to lpl2 and 3pl3 have been exchanged, as have the segments
distal to 3q25 and 9q22.

2.4.4.5 Suppressed Centromeres and Terminal Rearrangements

The increased precision of chromosome banding techniques has allowed the identification of a
special class of chromosome rearrangement in which one rearranged product contains two
centromere regions. However, only one, presumably the active

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one, appears as a primary constriction, while the other is apparently suppressed. Such
chromosomes may result from breakage and asymmetrical exchange or from the end-to-end
joining of two chromosomes (terminal rearrangement, ter rea). The chromosome carrying the
suppressed centromere is called a pseudodicentric, psu dic (similarly, pseudotricentric, psu tri,
etc.), and the segment with the presumptively active centromere is always written first. The term
cen is used only in the long form of the terminology to indicate the primary constriction, e.g.:

45, XX, ter rea(12;14)(pl3;p13)
45, XX, ter rea(12;14)(12qter→cen→12p13::14p13→14qter)

Cell with 45 chromosomes and a terminal rearrangement involving one chromosome 12 and one
chromosome 14. The abbreviation ter rea indicates that the abnormal chromosome has only one
active centromere and is not an ordinary dicentric chromosome. The centromere of the
chromosome mentioned first, i.e., chromosome 12, is the active one. In the detailed description,
the active centromere is indicated by cen.

47, XX, + psu dic(15)t(15;13)(ql2;ql2)
47, XX,+psu dic(15)t(15;13)(15pter→cen→15ql2::13ql2→13pter)

Cell with 47 chromosomes and an extra, pseudodicentric chromosome originating from a
translocation between one chromosome 15 and one 13. As seen from the detailed description, the
active centromere belongs to chromosome 15, which, therefore, is described before chromosome
13.

Marker Chromosomes (see also Section 4.2.3)

Any morphologically distinguishable abnormal chromosome that cannot be fully characterized
may be designated by the symbol mar. If part of the chromosome can be identified, the short
system can be used to describe the karyotype. A question mark (?) designates the unidentified segment. For example, 46, XX, t(12;?)

2.4.4.7 Derivative and Recombinant Chromosomes

Derivative chromosomes: A derivative chromosome in one of the structurally rearranged chromosomes generated by a rearrangement involving two or more chromosomes. The term is necessary if the short system is to be used (1) because a designation in this system symbolizes the rearrangement as such and not the chromosomes generated by the rearrangement, although these can nevertheless be identified from the symbolic designation and (2) in order to designate unbalanced karyotypes among offspring of structural heterozygotes which may include any one, or any combination, of the derivative chromosomes.

Recombinant chromosomes: A recombinant chromosome is a structurally rearranged chromosome with a new segmental composition resulting from meiotic crossing-over between a displaced segment and its normally located counterpart in certain types of structural heterozygotes.

Whereas derivative chromosomes are products of the original rearrangement and segregate at meiosis without further change, recombinant chromosomes arise de novo during gametogenesis in appropriate structural heterozygotes as predictable consequences of crossing-over in a displaced segment.

Derivative chromosomes are designated by the abbreviation der and recombinant chromosomes by rec. In both cases, the chromosome number is specified within parentheses immediately following the appropriate abbreviation. The chromosome number used is that which indicates the origin of the centromere of the particular derivative or recombinant chromosome.

As an illustration of the way derivative chromosomes can be expressed, a balanced reciprocal translocation between chromosomes 2 and 5, 46, XX, t(2;5)(q21;q31), has been assumed and is represented by the pachytene diagram in Fig. 7. The derivative chromosomes from such a translocation would be designated der(2) and der(5). Table 4 gives the possible unbalanced gametes resulting from adjacent-1 and adjacent-2 disjunctions and also from 4 of the 12 possible 3:1 disjunctions, together with the

Normal Chromosomes and Constitutional Aberrations

Fig. 7. Pachytene diagram of a t(2;5)(q21;q31) reciprocal translocation heterozygote used to specify the disjunctional possibilities and derivative chromosome combinations given in Table 3. Letters A, B, C, and D designate whole segments extending from chromosome ends (telomeres) to break points. Bands delimiting break points, only approximately to size, are shown.

Recommended designations of the karyotypes resulting from syngamy between each unbalanced gametic type and a normal gamete. The full karyotype designation need be written only once in any given publication and then can be abbreviated. A suggested abbreviation for the first designated karyotype in Table 4, for example, would be 46, XX, der(5)mat.

Recombinant chromosomes are most likely to originate from crossing-over in inversion or insertion heterozygotes. To exemplify the method of designating these chromosomes, a pericentric inversion of chromosome 2, 46, XX, inv(2)(p21q31), has been assumed and is shown diagrammatically in Fig. 8. In this case, crossing-over results in a duplication (dup) of 2p in one recombinant chromosome and of 2q in the other. The respective karyotype could be recorded as:
46, XX, rec(2), dup p, inv(2)(p21q31) and 46, XX, rec(2), dup q, inv(2)(p21q31), specifying, in the first example, a duplication from 2pter to 2p21 and a deletion from 2q31 to 2qter and, in the second example, a duplication from 2q31 to 2qter and a deletion from 2pter to 2p21.

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Table 4

Designation of unbalanced karyotypes. Use of der symbol to designate unbalanced karyotypes derived by segregation in a reciprocal translocation heterozygote. Based on the pachytene diagram in Fig. 7.

Disjunction (segregation)

Unbalanced gamete

Karyotype of zygote resulting from an unbalanced gamete fertilized by a normal gamete

| Adjacent- | ABCB | AD CD |
| Adjacent- | ABAD | CDBC | ABAB | AD AD | CBCB | CD CD |
| 3:12 | AB CB | AD | CB CD | AB | CD AD | CB | AD AB | CD |

46, XX, – 5, + der(5), t(2;5)(q21 ;q31)mat 46, XY, – 2, + der(2), t(2;5)(q21 ;q31)mat
46, XY, -5, + der(2), t(2;5)(q21;q31)mat
46, XY, – 2, + der(5), t(2;5)(q21 ;q31)mat
46, XX, + 2,-5
46, XY, – 2, - 5, + der(2), + der(2), t(2;5)(q21 ;q31)mat
46, XY, – 2, - 5, + der(5), + der(5), t(2;5)(q21 ;q31)mat
46, XX, -2, + 5
47, XX, + der(5), t(2;5)(q21;q31)mat
45, XY, – 2, – 5, + der(2), t(2;5)(q21 ;q31)mat
47, XX, – 2, + der(2), + der(5), t(2;5)(q21 ;q31)mat
45, XY, -5
47, XY, + der(2), t(2;5)(q21 ;q31)mat
45, XX,-2, -5, + der(5), t(2;5)(q21;q31)mat
47, XX, – 5, + der(2), + der(5), t(2;5)(q21 ;q31)mat
45, XX, -2

Adjacent-2 disjunction minimally results in the first two unbalanced gametic types shown (AB AD, CB CD). Crossing-over in the interstitial segments between centromeres and points of exchange is necessary for the origin of the remaining four types.
A further eight segregational types can occur at All if there is crossing-over in the interstitial segments, making a total of 12 types of gametes with three chromosomes derived from the translocation quadrivalent.

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INVERSION

In(2)(p21q31)
rec(2)dup p,inv(2)(p21q31)
p22 p16
q24 i q32

RECOMBINANTS

(Crossing over H) gi —o as shown) q324 * p16
q24 i q32
rec(2)dup q,inv(2)(p21q31)

Fig. 8. Designation of recombinant chromosomes in a pericentric inversion of chromosome No. 2 with break points in 2p21 and 2q31.

3.1 VARIATION OF SECONDARY CONSTRICCTIONS OR SATELLITES IN NON-BANDED CHROMOSOMES

Increases or decreases in the length of secondary constrictions, in that of negatively staining regions, or in the size of satellites should be distinguished from increases or decreases in arm length owing to other structural alterations by placing the symbol h or s between the symbol for the arm and the + or — sign, e.g.:

46, XY,16qh+ Male karyotype with 46 chromosomes, showing an increase in length of the secondary constriction on the long arm of chromosome 16
46, XY,21s+ Male karyotype with 46 chromosomes showing an increase in the size of the satellite on chromosome 21

3.2 VARIATION IN HETEROMORPHIC REGIONS IN BANDED CHROMOSOMES

3.2.1 Short Terminology

Duplication chromosome structures are indicated by repeating the appropriate designation. Thus, 46, XX,21pss would describe the karyotype of a female in which one chromosome 21 has double satellites on the short arm. While 46, XX,18ps indicates a chromosome 18 with satellites on the short arms, 46, XX,21psqs designates a chromosome 21 with satellites on the long and short arms.
The short form, such as lqh+ and 13s+, may still be used, but, when appropriate, a description of the technique used should be included, e.g., lqh+(CBG),21s(QFQ) (Table 5). Any code should be defined in the text of the publication in which it is first used. Examples of the use of this code are given in Section 3.2.2.

Table 5

Examples of code to describe banding techniques. In this one-, two- or three-letter code, the first letter denotes the type of banding, the second letter the general technique, and the third letter the stain. Some examples are given below.

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>O</td>
<td>O-bands</td>
</tr>
<tr>
<td>OF</td>
<td>O-bands by fluorescence</td>
</tr>
<tr>
<td>OFQ</td>
<td>O-bands by fluorescence using quinacrine</td>
</tr>
<tr>
<td>QFH</td>
<td>Q-bands by fluorescence using Hoechst</td>
</tr>
<tr>
<td>G</td>
<td>G-bands</td>
</tr>
<tr>
<td>GT</td>
<td>G-bands by trypsin</td>
</tr>
<tr>
<td>GTG</td>
<td>G-bands by trypsin using Giemsa</td>
</tr>
<tr>
<td>GAG</td>
<td>G-bands by acetic saline using Giemsa</td>
</tr>
<tr>
<td>C</td>
<td>C-bands</td>
</tr>
<tr>
<td>CB</td>
<td>C-bands by barium hydroxide</td>
</tr>
<tr>
<td>CBG</td>
<td>C-bands by barium hydroxide using</td>
</tr>
<tr>
<td>R</td>
<td>R-bands</td>
</tr>
<tr>
<td>RF</td>
<td>R-bands by fluorescence</td>
</tr>
<tr>
<td>RFA</td>
<td>R-bands by fluorescence using acridine</td>
</tr>
<tr>
<td>RH</td>
<td>R-bands by heating</td>
</tr>
<tr>
<td>RHG</td>
<td>R-bands by heating using Giemsa</td>
</tr>
<tr>
<td>RB</td>
<td>R-bands by BrdU</td>
</tr>
<tr>
<td>RGB</td>
<td>R-bands by BrdU using Giemsa</td>
</tr>
<tr>
<td>RBA</td>
<td>R-bands by BrdU using acridine orange</td>
</tr>
</tbody>
</table>

Complete Description

Heteromorphic chromosomes can be described if the term variable, abbreviated to var, is used before the chromosome number, e.g., var(13). Additional information regarding the variable region can then be conveyed by means of symbols set within brackets in the following order: (1) The location of the variable structure on the chromosome by either band numbers or code letters, such as cen, h, s etc. This is followed by a comma. (2) The banding technique used (see examples in Table 5). (3) A numerical designation for the size and staining intensity of the variable region, with higher numbers indicating greater size or staining intensity. Such numerical designations must be clearly defined (examples are given in Table 6). A zero indicates that size or intensity is not observed.

Table 6

Examples of how size and intensity descriptions can be expressed numerically.

<table>
<thead>
<tr>
<th>Size</th>
<th>Intensity (Paris Conference, 1971)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Q-banding</td>
<td></td>
</tr>
<tr>
<td>1 Very small</td>
<td>1 Negative (no or almost no</td>
</tr>
<tr>
<td>2 Small</td>
<td>2 Pale (as on distal ln)</td>
</tr>
<tr>
<td>3 Intermediate</td>
<td>3 Medium (as the two broad bands on</td>
</tr>
<tr>
<td>4 Large</td>
<td>4 Intense (as the distal half of 13q)</td>
</tr>
</tbody>
</table>
Very large: C-banding
Very small: 0 No quantitation of intensity
Small
Intermediate
Large
Very large

The definitions of "small," "large," etc., should be clearly presented in specific publications where the terminology is employed. A zero (0) may be used in any instance where quantitation is not used.

If quantitation of intensity of C-bands becomes possible, then an analogous series of definitions will be necessary.

Intensity was not quantitated. The number of digits used to describe size must equal the number of digits used to describe intensity.

When several techniques are used, each description should be separated by a comma. Their order is arbitrary. If more than one variable structure is present on the same homolog, each should be described in the same way, with parentheses, rather than a comma, used to separate the descriptions.

If the same variant appears on more than one homolog, an equal sign (=) followed by a number, e.g., var(13=2), can be used to designate the number of chromosomes that conform to the initial var description. The parental origin of a chromosome can be indicated by inserting pat or mat after the last parenthesis.

When more than one variable chromosome of a complement are described, the chromosomes in question are listed in descending order of chromosome size, the terms relating to each chromosome being separated by a comma. Bands on a given chromosome should be listed sequentially from the centromere outward, with those bands in the short arm listed first and those in the long arm last, e.g.:

46, XY, var(3)(cen, Q35)
Chromosome 3 with a centromeric region that, when Q-banded, is of intermediate size and fluoresces brilliantly

46, XY, var(13 = 2)(pl3, Q35)
Two chromosomes 13 with satellites (pl3) that, when Q-banded, are of intermediate size and fluoresce brilliantly

46, XY, var(13)(pl3, QFQ55, CBG50)(qll, QFQ35, CBG30)
One chromosome 13 with very large satellites (pl3) seen after both Q- and C-banding. These are brilliant after Q-banding, but C-banding intensity was not determined. In addition, band qll, when Q-banded, is of intermediate size and fluoresces brilliantly, and when C-banded, it is likewise intermediate in size. C-banding intensity was not determined.

46, XY, var(13)(pl3, QFQ45, CBG40)(qll, QFQ35, CBG30)
One chromosome 13 with large satellites (pl3) seen after both Q-banding by quinacrine fluorescence and C-banding by barium hydroxide pretreatment followed by Giemsa staining. These are brilliant after Q-banding, but C-banding intensity was not determined. In addition, band qll, when Q-banded, is of intermediate size and fluoresces brilliantly, and when C-banded, it is likewise intermediate in size. C-banding intensity was not determined.

46, XY, var(13)(qll, QFQ55), var(21)(pl3, QFQ44), var(22)(pl3, QFQ35)
Three variant chromosomes seen after Q-banding by quinacrine fluorescence

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47, XY, + 21, var(21)(pl3, Q12), var(21 = 2)(pl3, Q54)mat
Male with 47 chromosomes and trisomy 21. One chromosome 21 has very small satellites of pale intensity after Q-banding. The two remaining chromosomes 21 are identical, with very large and intensely fluorescent satellites, and both are of maternal origin.

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4 Nomenclature for Acquired Chromosome Aberrations
The purpose of this section is to provide a nomenclature for those acquired aberrations not already adequately described by the existing terminology for constitutional aberrations. Since many induced aberrations are scored on unbanded material, recommendations are given first for unbanded preparations; these are followed by brief recommendations for banded material. The aim is a simple, rather than comprehensive, nomenclature, and it is recognized that the proposals will not cover all situations. Its use, therefore, is recommended wherever practicable.

4.1 CHROMATID ABERRATIONS 4.1.1 Non-Banded Preparations
A chromatid (ct) aberration involves only one chromatid in a chromosome at a given locus. A chromatid gap (ctg) is a non-staining region (achromatic lesion) of a single chromatid in which there is minimal misalignment of the chromatid (Fig. 9a, b).
A chromatid break (ctb) is a discontinuity of a single chromatid in which there is a clear misalignment of one of the chromatids (Fig. 9c, d).
A chromatid exchange (cte) is the result of two or more chromatid lesions1 and the subsequent rearrangement of chromatid material (Fig. 10a-c). Exchanges may be between chromatids of different chromosomes (interchanges) or between or within chromatids of one chromosome (intrachanges). In the case of interchanges, it will generally be sufficient to indicate whether the configuration is triradial (tr) when there are three arms to the pattern, quadriradial (qr) when there are four, or complex (ex) when there are more than four. The number of centromeres might be indicated within parentheses (1cen, 2cen, etc.). When necessary, exchanges may be classified in more detail. Asymmetrical exchanges

1 Often referred to as a break points, but “lesion” is used here to denote an abnormality induced in the chromosome which may lead to a breakage event.

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(2cen), triradial, and complete. b. Chromatid exchange (cte): dicentric (2cen), quadriradial, and complete.
In complete exchanges all the broken ends are rejoined, but not in incomplete ones. In asymmetrical
exchanges, the incompleteness may be proximal when the broken ends nearest the centromere
are not rejoined or distal when the ends furthest from the centromere are not rejoined. Intra-arm
events include duplications, deletions, paracentric inversions, and isochromatid breaks showing
sister reunion. It should be noted that these terms are only descriptive and do not imply
knowledge of the origin of the aberrations.

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A sister chromatid exchange (see), when equal, complete, and symmetrical, is ordinarily
detectable only by autoradiography or by special staining methods. It results from the
interchange of homologous segments between two chromatids of one chromosome (Fig. 9h, i).
The abbreviation see is commonly used and should therefore be retained.

Banded Preparations
Some chromatid aberrations can be defined more precisely or can be recognized with certainty
only in banded preparations; e.g., a chromatid deletion (ct del) is the absence of a banded
sequence from only one of the two chromatids of a single chromosome. A chromatid inversion
(ct inv) is the reversal of a banded sequence of only one of the two chromatids of a single
chromosome. Both are subclasses of chromatid exchanges (cte).
Where it is desired to specify the location of a chromatid aberration, the appropriate symbol can
be followed by the band designation, e.g.:
Chromatid gap in chromosome 4 at band 4q25
Chromatid break in chromosome 4 at band 4q25
Chromatid exchange involving chromosomes 4 and 10 at bands 4q25 and 10q22, respectively
Chromatid deletion in chromosome 1 with absence of the band sequence lq2 to lq25
Chromatid inversion in chromosome 1 with reversal of the band sequence lq2 to lq25
Sister chromatid exchanges in chromosome 4 at bands 4q25 and 4q33
tctg(4)(q25)
tctb(4)(q25)
tcte(4;10)(q25;q22)
tct del(1)(q2q25) tct inv(1)(q2q25) sce(4)(q25q33)
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4.2 CHROMOSOME ABERRATIONS 4.2.1 Non-Banded Preparations
A chromosome (cs) aberration involves both chromatids of a single chromosome at the same
locus.
A chromosome gap (csg) is a non-staining region (achromatic lesion) at the same locus in both
chromatids of a single chromosome in which there is minimal misalignment of the chromatids
(Fig. 9e, f). The term “chromosome gap” is synonymous with “isolocus gap” and “isochromatid
gap.”
A chromosome break (csb) is a discontinuity at the same locus in both chromatids of a single
chromosome, giving rise to an acentric fragment and an abnormal monocentric chromosome
(Fig. 9g). This fragment is therefore a particular type of acentric fragment (ace), and csb should
be used only when the banding pattern or morphology indicates that the fragment is the result of
a single event. The terms “isolocus break” and “isochromatid break” are used to describe a chromatid-type aberration showing sister reunion at one or both ends (see cïe).

A chromosome exchange (cse) is the result of two or more chromosome lesions and the subsequent relocation of both chromatids of a single chromosome to a new position on the same or on another chromosome. It may be symmetrical (e.g., reciprocal translocation) or asymmetrical (e.g., dicentric formation). In non-banded preparations the terminology is exactly the same as for constitutional aberrations (Section 2.4.3).

A minute (min) is an acentric fragment smaller than the width of a single chromatid. It may be single or double. In the special situation found in tumor cells, where multiple double minutes are present, the abbreviation dmin may be used.

Pulverization (pvz) indicates a situation where a cell contains both chromatid and/or chromosome gaps and breaks which are not normally associated with exchanges and are present in such numbers that they cannot be enumerated. Occasionally, one or more chromosomes in a cell are pulverized while the remaining chromosomes are of normal morphology; e.g., pvz(l) is a pulverized chromosome 1.

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Premature chromosome condensation (pec) occurs when an interphase nucleus is prematurely induced to enter mitosis. This usually takes place in the cytoplasm of another cell, the nucleus of which is in a more advanced state of mitosis. A pec may involve an unreplicated G1 or a G2 nucleus. The chromatin of S-phase nuclei undergoing pec often appears to be pulverized.

Banded Preparations

When banded preparations allow adequate identification of chromosome segments or other chromosome aberrations, the nomenclature system recommended throughout this report can be used. When not, the observations can be described in words.

Marker Chromosomes (see also Section 2.4.4.6)

A marker chromosome (mar) is a structurally abnormal chromosome, banded or unbanded. When the banding pattern can be recognized, it can be adequately described by the standard nomenclature. When markers in different cells are known or thought to be the same, they may be indicated by a number after mar, e.g., mar1, mar2.

A number before mar indicates the number of markers; e.g., +2mar1 indicates two markers 1, while +13mar indicates 13 different unidentified or unique markers. The term Philadelphia (Ph1) chromosome is retained to describe the acquired deletion of the long arm of one G-group chromosome observed in the leukemic cells of the majority of cases of chronic myeloid leukemia, e.g., 46, XY, Ph1, where the chromosomes are not banded. Note that this does not define the translocation of which the Ph1 chromosome is one component and which should be described precisely by the standard nomenclature, e.g., 46, XY, t(9;22)(q34;q11).

Complex Chromosome Rearrangements

The short system of the standard nomenclature may give rise to ambiguities in describing chromosomes derived from some complex

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22 L6 22 U0

9 22 9 3

Fig. 11. Example of a complex rearrangement involving two homologous chromosomes; see text for description.
rearrangements, especially when two homologous chromosomes are involved. However, the
detailed system (Section 2.4.4.4) will clarify almost all situations.
The complex translocation shown in Fig. 11 may be described as follows:
der(9)t(3;9;22)(3pter→3pl4::9p21→9q32::9q21→9q34::22q11→22qter) and is a derivative
chromosome formed by three exchange events involving one chromosome 3, both chromosomes
9, and one chromosome 22. The presence of two chromosomes 9 in the bracket, which specifies
the chromosomes involved, will indicate that both homologs are represented in the abnormal
chromosome. The position of the breaks will in most cases distinguish between the homologs. If
this is not sufficient to distinguish between homologs, 9mat and 9pat may be used if this is
known from a study of heteromorphisms, but if the homologs
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cannot be distinguished, one of the numerals should be doubly underlined (9 and 9). In some
cases it will be necessary to illustrate the rearrangement and describe it in words to ensure
complete clarity.
Exchanges at the Centromere
Where an abnormal monocentric chromosome has one arm derived from one chromosome and
the other from a different chromosome, the aberration may be designated by assuming trans-
location at the centromeres, e.g., 46, XX, — 15, + t(lpl5p), but only the long form will make it
completely clear (see also “Whole-arm Translocations” in Section 2.4.4.4).
Chromosome Number
The chromosome count in a given cell should include all centric structures present in that cell
regardless of the number of centromeres in each structure; e.g., a dicentric chromosome is one
structure. Acentric fragments (ace) are not included in the count. Note also that when a tri-
radial or other complex chromatid rearrangement is present, it counts only as one structure regardless
of the number of centromeres.
When a sex chromosome is either missing or present in addition to the normal complement as an
acquired aberration, this should be indicated by adding a — or + sign followed by X or Y (see
also Section 4.5), e.g.:
45, X, — Y An acquired aberration in an individual with a 46, XY chromosome constitution in
which the cell in question lacks the Y chromosome
47, XX,+X An acquired aberration in an individual with a 46, XX chromosome constitution in
which the cell in question has an additional X chromosome
Description of a single abnormal cell is often not required, but a few examples may help to make
the terminology clear. Normally,
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the numerical abnormalities should be put first, in order of chromosome number (regardless of
sign), followed, in turn, by structural chromosome rearrangements, other chromosome
aberrations, and chromatid aberrations, e.g.:
45, XY,-B,-C,+D,-G, + dic(B;C), + ace,5ctg
Derived from a 46, XY cell, this abnormal cell lacks one G chromosome, has an additional D
chromosome, and has a dicentric composed of the missing B and C chromosomes. In addition,
there is an acentric fragment and five chromatid gaps.
45, XX, − B, − D, − F,+mar 1, csg, tr(2cen), ctb
Derived from a 46, XX cell, this abnormal cell lacks one B chromosome, one D chromosome, and one F chromosome; there is a marker, a chromosome gap, a dicentric triradial chromatid exchange, and a chromatid break. The marker chromosome replaces one of the missing chromosomes, and the triradial is composed of the two other missing chromosomes.

4.3 SCORING OF ABERRATIONS

In the scoring of aberrations, the main types are ctg, ctb, cte, csg, csb, ace, min, r, di, tr, der, and mar, and reports should, where possible, give the data under these headings. It is recognized, however, that aberrations are frequently grouped to give adequate numbers for statistical analysis or for some other reason. When this is done, it should be indicated how the groupings relate to the aberrations listed above, e.g.:

- Chromatid aberrations (ctg, ctb, cte) fragments (deletions) (csb, ace, min)
- Asymmetric aberrations (ace, di, tr)

1 Designated as “Cu” by Buckton and Pike (1964).

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The data should not be presented as deduced breakages per cell but in such a manner that it is possible to calculate the number of aberrations per cell.

CELL POPULATIONS WITH ACQUIRED ABNORMALITIES

A clone is a population of cells derived from a single progenitor cell. It is common practice to infer a clonal origin when a number of cells have the same or related abnormal chromosome complement. A clone is not necessarily homogeneous. When the term “clone” is used in this sense, it should be defined by the author since the precise definition will depend on both the number of cells examined and on the nature of the aberration involved. It will always mean at least two cells with the same aberration; e.g., two cells with the same additional complex marker among 25 analyzed cells would be an acceptable clone, but two 45, X cells among 100 normal female cells would not. Thus, in some cases, especially with hypodiploid cells, three or even more cells will be necessary to constitute a clone, depending on the number of cells analyzed.

TUMOR CELL POPULATIONS

The individual chromosomes of tumor cell populations can all be described by the standard nomenclature. There is a need, however, to clarify the terminology of populations of tumor cells. Modal number (mn) is the most common chromosome number in a tumor cell population at the time that it is observed in a direct preparation or following short-term culture. (Short-term culture includes incubation for a few days without mitogens.) When long-term cultured cells are used, this should be indicated by the words in vitro in front of the term modal number. The modal number may be described as near diploid (2n ± ) when it is approximately diploid but the mode is not sharp. The modal number will be hypodiploid (2n—) when the mode is less than 46 chromosomes and hyperdiploid (2n+) when it is more than 46 chromosomes but, in either case, there is no sharp mode at a particular chromosome number.

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Modal numbers in the triploid or tetraploid range or near but not equal to any other multiple of the haploid number and which cannot be given as a precise number of chromosomes may be expressed as 3n ± (near triploid), 3n— (hypotriploid), 3n+ (hypertriploid), 4n ± (near tetraploid), 4n — (hypotetraploid), 4n+ (hypertetraploid), and so on.
The distinction between 2n+ and 3n— would be that in the former case the majority of the counts lie below 2n + (n/2), and in the latter the majority of the counts lie above 2n+(n/2). A similar distinction can be made between 3n+ and 4n —, and so on.

Pseudodiploid, pseudotriploid, etc., are used to describe a cell which has the number of chromosomes equal to a ploidy level but is abnormal because of the presence of acquired numerical and/or structural aberrations.

Stem-line (si) indicates the most frequent chromosome constitution of a tumor cell population at the time that it is observed in a direct preparation or following short-term culture. Again, the term in vitro should be used to indicate that long-term cultured cells were studied. All other lines are termed side-lines (sdl) or sub-lines. If there are a number of lines that appear to be related, these could be referred to as a clone (see above); e.g., in Table 7 the stem-line and side-line variation always involves a clone, 46, XX, t(9;22).

If more than one side-line is present, these should be referred to as sdl 1, sdl 2, and so on. The sdl number does not indicate the frequency of the side-line. The same designation for a side-line should be used for the same side-line in sequential observations. Where a side-line is found to have split into two separate side-lines at a second or subsequent observation, this should be indicated by retaining the original number for the unchanged side-line and giving a new sdl number to the new line (see the examples in Table 7).

In writing down the chromosome constitution of a tumor, both the number of cells counted and the number of cells analyzed should be given. The chromosome constitution of a tumor will consist of two series of symbols separated by a period (.). The first series will give the numbers of cells in each category with a different number of chromosomes. The second series will give the numbers of cells in each category with a different chromosome constitution. Thus 46 = 5/49 = 10/52 = 4. 46, XY = 2/49, XY, + 1.

### Acquired Chromosome Aberrations

Table 7

Sample descriptions of sequential observations in a single case of chronic myeloid leukemia. The stem-line in observation 1 is a line with a simple translocation resulting in the Phi chromosome. This line is succeeded by another stem-line in observation 3, becomes side-line 4, and is not seen in subsequent observations. The line with 47 chromosomes and an extra chromosome 8 is present throughout as side-line 2, except for observation 3, where it has become the stem-line. The side-lines 1 and 3 apparently disappear without further evolution, but side-line 5, and the new stem-line in observation 5, could have been derived from side-line 2.

<table>
<thead>
<tr>
<th>Observation</th>
<th>si</th>
<th>sdl1</th>
<th>sdl2</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>46, XX, t(9;22)</td>
<td>47, XX, 22a, + 22q-</td>
<td>47, XX, 22a, + 8</td>
</tr>
<tr>
<td>Observation</td>
<td>si</td>
<td>sdl1</td>
<td>sdl2</td>
</tr>
<tr>
<td></td>
<td>46, XX, 22</td>
<td>47, XX, 22a, + 8</td>
<td></td>
</tr>
<tr>
<td>Observation</td>
<td>si</td>
<td>sdl1</td>
<td>sdl2</td>
</tr>
<tr>
<td></td>
<td>47, XX, 22a, + 9, + 10</td>
<td>46, XX, 22a, + 8</td>
<td></td>
</tr>
<tr>
<td>Observation</td>
<td>si</td>
<td>sdl1</td>
<td>sdl2</td>
</tr>
<tr>
<td></td>
<td>47, XX, 22a, + 8</td>
<td>47, XX, 22a, + 8</td>
<td></td>
</tr>
<tr>
<td>Observation</td>
<td>si</td>
<td>sdl1</td>
<td>sdl2</td>
</tr>
<tr>
<td></td>
<td>48, XX, 22a, + 9, + 10</td>
<td>46, XX, 22a, + 8</td>
<td></td>
</tr>
<tr>
<td>Observation</td>
<td>si</td>
<td>sdl1</td>
<td>sdl2</td>
</tr>
<tr>
<td></td>
<td>48, XX, 22a, + 8, + 8</td>
<td>47, XX, 22a, + 8</td>
<td></td>
</tr>
</tbody>
</table>
Observation

sd
48, XX, (22), + 9, + 10
sd
49, XX, (22), + 8, + 8, - 17, + 10
sd
47, XX, (22), + 8
sd
48, XX, (22), + 8, + 8

+ 8, + 12 = 9/49, XY, + X, + 1, + 7 = 1/52, XY, + X, + X, + 1, + 7, + 22, + 22 = 4 indicates a tumor
in which a total of 19 cells were counted. The number of cells with 46 chromosomes is 5, the
number with 49 chromosomes is 10, and the number with 52 chromosomes is 4. Of the cells with
46 chromosomes, 2 had been analyzed, and both have a normal male karyotype. Of the cells with
49 chromosomes, 10 had been analyzed; 9 of these have an additional chromosome 1, an
additional 8, and an additional 12, while 1 of them has an additional X chromosome, an
additional 1, and an additional 7. Of the cells with 52 chromosomes, all 4 had been analyzed, and
all have two additional X chromosomes, one
additional 1, one additional 7, and two additional 22’s (cf. nomenclature for mosaics, Section
2.4.2.2).

If there is no restriction on space, some authors may prefer to put each item of information on a
separate line.

Determination of the stem-line requires consideration of the number of cells counted and
analyzed as well as the number of cells with a given karyotype. Thus, for the tumor 46 = 5/47 =
35/48 = 10. 46, XY = 3/47, XY, + 8 = 6/47, XY, + X = 4/48, XY, + 8, + 9 = 1/48, XY, + 21, +
21 = 1, the calculation is as follows:

Out of 50 cells counted, 5 had 46 chromosomes; of these 5, all three of the three cells analyzed
had a 46, XY karyotype. Assuming that the sampling is random,
46, XY 5/50 X 3/3 X 100 % = 10 %

Similarly,
47, XY, + 8 35/50 X 6/10 X 100 % = 42 %
47, XY, + X 35/50 X 4/10 X 100 % = 28 %
48, XY, + 8, + 9 40/50 X 1/2 X 100 % = 10 %
48, XY, + 21, + 21 40/50 X 1/2 X 100 % = 10 %

Therefore, the stem-line is 47, XY, + 8.

It is not necessary to describe the analysis of all side-lines and unique cells in the description of a
tumor since, in some cases, the number of separate karyotypes may be very large. It will be
implicit in the description of a tumor, however, that those karyotypes not included are less
numerous than those that are included.

When variability is very great, the chromosome constitution of the tumor could be reported
simply as, e.g., 2n = 37/4n = 3/8n = 1 (s/k, XX, + 8 = 13) meaning that 37 near-diploid,
three hypotetraploid, and one hypo-octaploid cells have been counted, and the stem-line is
represented by 13 cells with an extra chromosome 8.

There will be some situations where it will be simpler to describe the chromosome constitution
of a tumor in words than to express it as a complex formula, and this should be done when
appropriate.

Human Meiotic Chromosomes

Nomenclature for Human Meiotic Chromosomes

During late prophase-first metaphase, the bivalents may be grouped by size, and bivalent 9 can
sometimes be distinguished by its secondary constriction. At these stages, the Q- and C-staining
methods are particularly informative. The autosomal bivalents generally show the same Q-band
patterns as somatic chromosomes. The C-staining method reveals the centromere position, thus allowing identification of the bivalents in accordance with the conventionally stained somatic chromosomes. There are, however, minor differences in the C-band patterns between the bivalents and mitotic chromosomes. When the Q- and C-staining methods are used consecutively, further distinction of the bivalents is possible. Measurements of the relative length of orcein-stained bivalents, previously identified by these special techniques, are in good agreement with corresponding mitotic measurements (Appendix 3). Chiasma frequencies have been determined for individual bivalents (Appendix 3). The Y chromosome can be identified at all meiotic stages by the intense fluorescence of its long arm. Both the Q- and C-staining methods have revealed that the short arm of the Y is associated with the short arm of the X in the first meiotic metaphase.

**TERMINOLOGY**

The abbreviations PI, MI, AI, MII, and AII are used to indicate the stage of meiosis, namely, prophase of the first division, first metaphase (including diakinesis), first anaphase, second metaphase, and second anaphase. This is followed by the total count of separate chromosomal elements. The sex chromosomes are then indicated by XY or XX when associated and as X,Y when separate. Any additional, missing, or abnormal element follows, with that element specified within parentheses and preceded by the Roman numeral I, II, III, or IV to indicate if it is a univalent, bivalent, trivalent, or quadrivalent, respectively. The absence of a particular element is indicated by a — sign. The + sign is used in first metaphase only when the additional chromosome is not included in a multivalent.

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The chromosomes involved in a rearrangement are listed numerically within parentheses and separated by a semicolon (;).

A more detailed description, for instance, of the chromosomal segments involved in a rearrangement may be included within parentheses using the standard nomenclature, with which this meiotic notation has been designed to conform. When necessary, use of the abbreviations mal and fern is recommended for male and female, respectively, and when a more detailed description of different premeiotic and meiotic stages is required, the following abbreviations may be used:

- spm: Spermatogonial metaphase
- oom: Oogonial metaphase
- lep: Leptotene
- zyg: Zygotene
- pac: Pachytene
- dip: Diplotene
- dit: Dictyate (not die, which is used for diakinesis)

The abbreviation xma is suggested for chiasma(ta). The total number of chiasmata in a cell can be designated by placing this abbreviation within parentheses, following it by an equal sign (=) and a two-digit number, e.g., (xma=52). In the case of a meiotic cell with a low number of chiasmata, a single digit should be preceded by a zero, e.g., (xma=09).

The number of chiasmata in a bivalent or multivalent or their arms may be indicated by a single digit, e.g., (xma=4).
Location of chiasmata can be indicated by the standard symbols p and q, supplemented by prx for proximal, med for medial, dis for distal, and ter for terminal. The band or region number can be used when such precise information is available. Chromosomes participating in a bivalent or multivalent are specified within parentheses after the Roman numeral that describes the bivalent (II) or the type of multivalent (III, IV, etc.). If the number of chiasmata within the multivalent is known, this is indicated within parentheses in consecutive order; i.e., the number of chiasmata between the first and second chromosome is given first, between the second and the third next, etc. The last figure then indicates the number of chiasmata between the last and first chromosome. If the number of chiasmata in non-interstitial and interstitial segments can be specified separately, these should be represented by a + sign. The number of chiasmata in the non-interstitial segment is written first, e.g., \((x\text{ma}=2 + 1)\), indicating two chiasmata in the non-interstitial and one in the interstitial segment. It is assumed that a careful description of the mitotic karyotype of the subject will be given separately.

Examples of Meiotic Nomenclature

**MI,23, XY**
A primary spermatocyte at diakinesis or metaphase I with 23 elements, including an XY bivalent

**MI,24, X, Y**
A primary spermatocyte at diakinesis or metaphase I with 24 elements, including X and Y univalents

**MI,23, XY, III(21)**
A primary spermatocyte with 23 elements from a male with trisomy 21. The three chromosomes 21 are represented by a trivalent.

**MI,24, XY, + I(21)**
A primary spermatocyte with 24 elements from a male with trisomy 21. The extra chromosome 21 is represented by a univalent.

**MI,22, XY, III(13q14q)**
A primary spermatocyte with 22 elements from a balanced t(13q14q) heterozygote. The t(13q14q) chromosome is represented by a trivalent.

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**MI,22, XY, IV(5;C)**
A primary spermatocyte with 22 elements from a male with a t(5;C) reciprocal translocation. The t(5;C) chromosome is represented by a quadrivalent.

**Spermatocyte in first metaphase with 23 structures, including an XY bivalent. The total number of chiasmata in the cell is 52, the association between the X and Y chromosomes being counted as one chiasma.**

**fern dia.,[I](2,2)(x\text{ma}=4)**
Oocyte in diakinesis in which bivalent 2 has four chiasmata.

**fern dia.,[I](2,2)(x\text{ma}=4)(p = 2, q = 2)**
Female diakinesis in which bivalent 2 has four chiasmata. The positions of the chiasmata are known. Thus \((x\text{ma}=4)(p = 2, q = 2)\) indicates that there are two chiasmata on the short arm and...
two on the long arm. More precise location of the chiasmata could then be indicated, e.g., by (xma = 4)(pter, pprx, qmed, qdis). Alternatively, if the chiasmata have been localized to specific regions, these could be indicated, e.g., by (xma=4)(pter, pl, q2, qter).

Male first metaphase with a trivalent composed of one chromosome 14, one 14q21q Robertsonian translocation chromosome, and one chromosome 21. There are three chiasmata, the positions of which have not been specified.

Spermatocyte in first metaphase with 23 structures, univalent X and Y chromosomes, and one trivalent composed of one chromosome 13, one 13q14q Robertsonian translocation chromosome, and one chromosome 14. There are two chiasmata between the normal chromosome 13 and the 13q14q translocation chromosome and one chiasma between the translocation chromosome and the normal chromosome 14. Altogether, there are 51 chiasmata in the cell.

Oocyte in diakinesis with a quadrivalent composed of two normal chromosomes and two derivative chromosomes of chromosomes 2 and 5, respectively. There are three chiasmata between chromosomes 2 and der(2), of which two are in the non-interstitial segment and one is in the interstitial segment. In addition, there is one chiasma between der(2) and chromosome 5, one in the non-interstitial and none in the interstitial segment between chromosome 5 and der(5), and finally one between der(5) and chromosome 2. The last chiasma indicates that the quadrivalent has a ring shape.

Spermatocyte in first metaphase with 24 structures, including univalent X and Y chromosomes, one trivalent, and one univalent. The trivalent is composed of one normal and one derivative chromosome 2, as well as one normal chromosome 5. This trivalent has a total of four chiasmata, the positions of which are not exactly known. The univalent is composed of one derivative chromosome 5. The total number of chiasmata in the cell is 51.

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Species Codes
A standardized shorthand method is recommended for designating interspecific animal or cell hybrids.

Designations for species. Only capital letters should be used. Subscripts, superscripts, or other symbols are undesirable. A three-letter abbreviation is chosen according to the following rules (see Table 8).

The first letter of the generic name and the first two letters of the species name are ordinarily selected; e.g., Homo sapiens becomes HSA; Equus caballus, ECA; Equus asinus, EAS; and Cricetulus griseus, CGR.

When a new abbreviation, selected according to the preceding rule, is indistinguishable from a previously accepted abbreviation, a unique abbreviation is obtained by selecting for the third letter of the abbreviation the second, third, or next most consecutive letter of the species name which provides a unique abbreviation; i.e., Mus musculus is identified as MMU and Macaca mulatta as MML. If no letter of the species name confers uniqueness, letters not available in the species name may be used. When subspecies exist, duplication may be avoided by use of the first
letter of the species name and the first letter of the subspecies name in conjunction with the initial letter of the generic name.

Interspecific animal hybrids. Mules, for example, are designated as (ECA*EAS), the hinney as (EAS*ECA). The asterisk indicates the product of a dual parentage. The species named first is the female parent; the second one is the male parent.

Interspecific somatic cell hybrids. Hybrids between human and Chinese hamster cells, for example, may be represented by the expression (HSA+CGR). The order of the species is arbitrary. The + sign is used to indicate the addition of genomes or parts of genomes.

Complex rearrangements occurring in somatic cell hybrids. These may be designated by combining the species abbreviation with the standard nomenclature for chromosome rearrangements. For compound interspecific chromosomes, the species abbreviations and chromosome abbreviations are always written in the same order. Thus, t(HSA + CGR)(1;9) indicates a translocation between a human chromosome 1 and a Chinese hamster chromosome 9.

Examples of Reciprocal Translocations

<table>
<thead>
<tr>
<th>Expression</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>t(HSA+CGR)(1;9)(q24;q12)</td>
<td>Breakage and reunion have occurred at band q24 of human chromosome 1 and band q12 of Chinese hamster chromosome 9 in a human-plus-Chinese hamster cell hybrid. Segments distal to these bands have been exchanged between the two chromosomes, and both the derivative chromosomes are present in the cell.</td>
</tr>
<tr>
<td>t(HSA+CGR)(1;9)(pter→lq24::q9l2→9qter; 9pter→q9l2::lq24→lqter)</td>
<td></td>
</tr>
</tbody>
</table>

If it is known that such a translocation has occurred, but only one derivative chromosome can be identified in the hybrid cell, it would be described, e.g., as:

der(HSA+CGR)(1;9)(pter→lq24::q9l2→9qter)

If only one of the chromosomes involved in a rearrangement can be recognized, the query sign (?) may be used to indicate additional chromosome material of unknown origin, e.g.:
t(HSA + CGR)(1;?) (pter→q24;?)

This indicates the presence of the pter→q24 segment of human chromosome 1 in a rearranged chromosome. The source of the remainder of that chromosome is not known.

The abbreviation rea may be used to describe a rearranged compound chromosome in a hybrid cell whose components can be identified, but the type of rearrangement which led to its formation is not known, e.g.:

rea(HSA + CGR)(1qter→lp22::7q32→7qter)

This indicates a rearranged compound chromosome consisting of the qter→p22 segment of human chromosome 1 joined to the q32→qter segment of Chinese hamster chromosome 7. When it is necessary to refer repeatedly to a number of different rearrangements, then the foregoing can be shortened to rea(a), rea(b), etc., provided these are fully defined.

Further changes in the rearranged chromosomes, such as occur in somatic cell hybrids, would be described as follows:
del rea(a)HSA(l)(q32q34)

This indicates that in the rearranged compound chromosome defined in the previous example, and subsequently labeled rea(a), an interstitial deletion of bands q32→q34 of the human chromosome 1 segment of the compound chromosome has occurred.

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Chromosomes of the Great Apes

Nomenclature and Presumptive Homologies for the Chromosomes of the Great Apes (Pongidae)

Extensive banding information is available on the karyotype of the great apes (Pongidae). It was considered that a nomenclature system based on chromosome morphology (Paris Conference, 1971, Supplement, 1975) did not conveniently express the phyllo-genetic relationships. A dual scheme is now proposed for the nomenclature of the karyotypes of the chimpanzee (Pan troglodytes, PTR), pygmy chimpanzee (Pan paniscus, PPA), gorilla (Gorilla gorilla, GGO), and orangutan (Pongo pygmaeus, PPY).

There are many similarities in the chromosome banding patterns of the species considered here when compared to those of the human karyotype. The double nomenclature provides a choice between a fixed reference system of numbering and a method for more directly indicating presumptive homologies.

**CONVENTIONAL NOMENCLATURE**

When referring to a particular chromosome, the species code is followed by an Arabic numeral which has been assigned on the basis of chromosome length and centromere position (Figs. 12-14). Homologies may be indicated by placing in parentheses the presumptive homologous chromosome and the code of the species to which it is being compared.

**PHYLOGENETIC NOMENCLATURE**

In this system, chromosomes are given Roman numerals corresponding to the presumptive human homologies based on banding patterns, as proposed in Table 8.

![Fig. 12. R-, Q-, and G-banded karyotypes of Pan troglodytes arranged according to length and centromere position. The banding pattern of Pan paniscus (PPA) is very similar to that of Pan troglodytes. For minor differences, see Dutrillaux et al. (1975) and Henderson et al. (1976).](http://example.com/fig12.jpg)
Chromosomes of the Great Apes

* [H] El 8°C
234
8
8 8 IIi
10 11 12
Os 7a 3
13 14 15 16
13? i& i.p
17 18 19 20
w wy y
21 22 23

13. Same as Fig. 12 but of Gorilla gorilla.

Table 8

Proposed chromosome band homologies of great apes and man, using conventional and phylogenetic nomenclatures.

<table>
<thead>
<tr>
<th>Homo sapiens</th>
<th>Pan troglodytes</th>
<th>Gorilla gorilla</th>
<th>Pongo pygmaeus</th>
</tr>
</thead>
<tbody>
<tr>
<td>(HSA)</td>
<td>(PTR)</td>
<td>(GGO)</td>
<td>(PPY)</td>
</tr>
<tr>
<td>1</td>
<td>1</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>2</td>
<td>11</td>
<td>12.13</td>
<td>12.11</td>
</tr>
<tr>
<td>3</td>
<td>III</td>
<td>III</td>
<td>III</td>
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<tr>
<td>4</td>
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<td>IV</td>
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<td>V</td>
<td>V</td>
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<tr>
<td>6</td>
<td>VI</td>
<td>VI</td>
<td>VI</td>
</tr>
<tr>
<td>7</td>
<td>VII</td>
<td>VII</td>
<td>VII</td>
</tr>
</tbody>
</table>

74. Same as Fig. 12 but of Pongo pygmaeus.
This banding homology has not been completely resolved.

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Examples

Conventional nomenclature: PPY 7 (HSA 10) Phylogenetic nomenclature: PPY X
In the conventional nomenclature, chromosome 7 of PPY would be indicated as above, and the presumptive homology between that chromosome and chromosome 10 of man can be shown by the component in parentheses. In the phylogenetic nomenclature, the Roman numeral X indicates the same homology. (Care should be taken to clearly distinguish between the Roman numeral X and the letter X.)

Conventional nomenclature: PPY 3 [HSA inv(4)(p15q21)] Phylogenetic nomenclature: PPY IV [HSA inv(4)(p15q21)]
This indicates that orangutan chromosome 3 (or IV) differs from chromosome 4 in man by a presumptive pericentric inversion with break points at 4p15 and 4q21 of the human chromosome.

COMPARATIVE BANDING DIAGRAMS

The bands represented diagrammatically in Figs. 15a-g have been identified with a variety of banding techniques and are numbered according to the landmark system adopted at the Paris Conference (1971). Where necessary, fub-band designations are used. The landmarks and band numbers have been selected to make comparisons with the human karyotype simpler, and the chromosomes have been arranged to show their presumptive homologies with the specific human chromosomes. The relative position and widths of some of the bands of the human chromosome diagram have been modified to emphasize the similarities between the chromosomes of man and the great apes. For the standard human karyotype, see Fig. 5.

In Figs. 15a-g, the Roman numeral at the left of each set of chromosomes represents the phylogenetic number, and the species code, as well as the same Arabic chromosome numbers used in

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Figs. 12-14, is given at the end of each long arm. The chromosome-specific band designations are at the left of each chromosome; the designations to the right are those of the presumptive human homologous bands. The following ape chromosomes, as depicted in the individual karyotypes (Figs. 12-14), have been inverted in Fig. 15 to maintain comparisons based on similarity of bands: chromosomes PTR 1 and PTR 13; GGO 1, GGO 12, and GGO 17; and PPY 1 and PPY 12.

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Diagrammatic representation of chromosome bands of Homo sapiens, Pan troglodytes, Gorilla gorilla, and Pongo pygmaeus, arranged from left to right, as observed with the R-, Q-, and G-staining techniques. The chromosomes have been arranged to show the similarities in banding pattern as described in the text. Open areas show negative or pale-staining Q and G bands, as well as positive R bands. Solid areas show positive Q and G bands and negative R bands. Cross-hatched areas depict variable bands. The conventional species numbers are given next to the chromosomes, and the phylogenetic Roman numbers are listed at the left; see text for further description. (Prepared by Dr. P. Pearson.)
<table>
<thead>
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<th>1</th>
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<tbody>
<tr>
<td>1</td>
<td>M1</td>
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<td>i</td>
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×

P12

q11 q21 q23 q25
q0 q21 q23 q25 q21 q23 q23 q25 q21 q23

XV

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<tr>
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XVII
1  2
1
2  2
HSA17
6
1 1
3
12
1
PTR19
9
1
1 2
3
π
5
66019

PPY19
XVIII

lie
1 1
2 2

HSA18

q12.1 q12.3
1
6
q21.2 q22
PTR17
1 5
6
2 2
66016
2H
q12 1
q123
```
Fig. 15f (see legend on p. 78).
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q21

2

HSA21
PTR22
GG022

q12

3© ©
1
1
V
1
2

1
V
2
1

1
V
2
4
w

30 (1
1
V
2
1
3

HSA22
PTR23
GG023

m

2
2
1
V
1
1
8
References


Standing Committee and Consultants 397 (85)

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10 Appendices

Appendix 1

Human somatic cell metaphase chromosome measurements: relative length (in percentage of the total haploid autosome length) and centromere index (length of short arm divided by total chromosome length × 100). Chromosomes stained with orcein or the Giemsa 9 method and pre-identified by Q-band patterns. Means and standard deviations are given.

<table>
<thead>
<tr>
<th>Crhomo-</th>
<th>Relative length</th>
<th>Centromere index</th>
</tr>
</thead>
<tbody>
<tr>
<td>No.</td>
<td>A</td>
<td>B</td>
</tr>
<tr>
<td>1</td>
<td>9.08</td>
<td>9.08 ±</td>
</tr>
<tr>
<td>2</td>
<td>8.45</td>
<td>8.17 ±</td>
</tr>
<tr>
<td>3</td>
<td>7.06</td>
<td>6.96 ±</td>
</tr>
<tr>
<td>4</td>
<td>6.55</td>
<td>6.62 ±</td>
</tr>
<tr>
<td>5</td>
<td>6.13</td>
<td>6.34 ±</td>
</tr>
<tr>
<td>6</td>
<td>5.84</td>
<td>6.19 ±</td>
</tr>
<tr>
<td>7</td>
<td>5.28</td>
<td>5.60 ±</td>
</tr>
<tr>
<td>X</td>
<td>5.80</td>
<td>5.45 ±</td>
</tr>
<tr>
<td>8</td>
<td>4.96</td>
<td>5.13 ±</td>
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<tr>
<td>9</td>
<td>4.83</td>
<td>4.81 ±</td>
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<td>10</td>
<td>4.68</td>
<td>4.66 ±</td>
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<td>11</td>
<td>4.63</td>
<td>4.70 ±</td>
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<tr>
<td>12</td>
<td>4.46</td>
<td>4.66 ±</td>
</tr>
<tr>
<td>13</td>
<td>3.64</td>
<td>3.22 ±</td>
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<tr>
<td>14</td>
<td>3.55</td>
<td>3.09 ±</td>
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<td>2.33</td>
<td>2.61 ±</td>
</tr>
<tr>
<td>21</td>
<td>1.83</td>
<td>1.34 ±</td>
</tr>
<tr>
<td>22</td>
<td>1.68</td>
<td>1.53 ±</td>
</tr>
<tr>
<td>Y</td>
<td>1.96</td>
<td>1.82 ±</td>
</tr>
</tbody>
</table>

A: Previous Denver-London data (not pre-identified by Q-staining method).

B: Data from 20 cells provided by Dr. P. Pearson. Cells stained with orcein. The short arms in groups D and G and in the Y were excluded.

C: Data from 10 cells provided by Drs. T. Caspersson, M. Hultén, J. Lindsten, and L. Zech. Cells stained with orcein.

D: Data from 95 cells provided by Drs. H. Lubs, T. Hostetter, and L. Ewing from 11 normal
subjects (6-10 cells per person). Average total length of chromosomes per cell: 176 µ. Cells
stained
with orcein or the Giemsa 9 technique. Cells in B, C, and D were measured from projected
negatives of metaphase cells. Standard deviations in samples B and C are based on the total
sample of measurements. Standard deviations in sample D are an average of the standard
deviations found in each of 11 subjects (6-10 cells per subject).

ISCN (1978)
Appendix 2
Diagrammatic representation of human chromosome bands as observed with the Q-, G-, and
R-staining methods; centromere representative of Q-staining method only (Paris Conference,
1971).

400 (88)

Appendix 2
401 (89)
Appendix 3

Human male meiotic chromosome measurements: relative length, centromere index, and number of chiasmata of individual bivalents identified with the Q-band technique, restained with orcein, and then identified again by C-banding. Data from 41 diakinetic cells from one control case with apparently normal spermatogenesis provided by Dr. M. Hultén. Data from 10 mitotic cells from two healthy subjects provided by Drs. T. Caspersson, M. Hultén, J. Lindsten, and L. Zech. Means and standard deviations are given.

<table>
<thead>
<tr>
<th>Bivalent number of bivalents</th>
<th>Relative length</th>
<th>Centromere index</th>
<th>Number of chiasmata</th>
<th>Relative number</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Meiotic Mitotic</td>
<td>Meiotic Mitotic</td>
<td>Meiotic Mitotic</td>
<td></td>
</tr>
<tr>
<td>1</td>
<td>33</td>
<td>9.06 ± 9.11</td>
<td>50.6 ± 48.6</td>
<td>2-5 3.90 ± 7.59</td>
</tr>
<tr>
<td>2</td>
<td>34</td>
<td>8.60 ± 8.61</td>
<td>38.8 ± 38.9</td>
<td>2-5 3.62 ± 7.11</td>
</tr>
<tr>
<td>3</td>
<td>33</td>
<td>6.86 ± 6.97</td>
<td>46.1 ± 47.3</td>
<td>2-4 2.92 ± 5.81</td>
</tr>
<tr>
<td>4</td>
<td>24</td>
<td>5.98 ± 6.49</td>
<td>27.0 ± 27.8</td>
<td>2-3 2.79 ± 5.48</td>
</tr>
<tr>
<td>5</td>
<td>26</td>
<td>6.36 ± 6.21</td>
<td>24.4 ± 26.8</td>
<td>2-4 2.85 ± 5.53</td>
</tr>
<tr>
<td>6</td>
<td>15</td>
<td>5.80 ± 6.07</td>
<td>34.1 ± 37.9</td>
<td>2-4 2.67 ± 5.04</td>
</tr>
<tr>
<td>7</td>
<td>19</td>
<td>5.64 ± 5.43</td>
<td>37.8 ± 37.0</td>
<td>2-4 2.74 ± 5.21</td>
</tr>
<tr>
<td>8</td>
<td>11</td>
<td>4.73 ± 4.94</td>
<td>30.7 ± 32.8</td>
<td>2-3 2.64 ± 4.99</td>
</tr>
<tr>
<td>9</td>
<td>34</td>
<td>54.57 ± 4.78</td>
<td>35.7 ± 32.7</td>
<td>2-3 2.41 ± 4.73</td>
</tr>
<tr>
<td>10</td>
<td>22</td>
<td>4.79 ± 4.80</td>
<td>32.1 ± 32.3</td>
<td>2-3 2.50 ± 4.96</td>
</tr>
<tr>
<td>11</td>
<td>24</td>
<td>4.53 ± 4.82</td>
<td>34.8 ± 40.5</td>
<td>2-3 2.21 ± 4.31</td>
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<tr>
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<td>17</td>
<td>4.73 ± 4.50</td>
<td>26.5 ± 27.4</td>
<td>2-3 2.71 ± 5.22</td>
</tr>
<tr>
<td>13</td>
<td>33</td>
<td>3.83 ± 3.87</td>
<td>9.5 ± 16.6</td>
<td>1-3 1.85 ± 3.63</td>
</tr>
<tr>
<td>14</td>
<td>17</td>
<td>3.73 ± 3.74</td>
<td>11.3 ± 18.4</td>
<td>1-3 1.88 ± 3.66</td>
</tr>
<tr>
<td>15</td>
<td>22</td>
<td>3.83 ± 3.30</td>
<td>12.0 ± 17.6</td>
<td>1-3 2.05 ± 4.03</td>
</tr>
<tr>
<td>16</td>
<td>25</td>
<td>3.10 ± 3.14</td>
<td>45.7 ± 42.5</td>
<td>2-3 2.16 ± 4.24</td>
</tr>
<tr>
<td>17</td>
<td>23</td>
<td>3.03 ± 2.97</td>
<td>31.1 ± 31.9</td>
<td>2-3 2.13 ± 4.14</td>
</tr>
<tr>
<td>18</td>
<td>24</td>
<td>2.72 ± 2.78</td>
<td>28.1 ± 26.6</td>
<td>1-2 1.92 ± 3.73</td>
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<tr>
<td>19</td>
<td>35</td>
<td>2.38 ± 2.46</td>
<td>45.7 ± 44.9</td>
<td>1-2 1.94 ± 3.82</td>
</tr>
<tr>
<td>20</td>
<td>35</td>
<td>2.45 ± 2.25</td>
<td>43.6 ± 45.6</td>
<td>1-2 2.00 ± 3.98</td>
</tr>
<tr>
<td>21</td>
<td>38</td>
<td>1.63 ± 1.70</td>
<td>20.5 ± 28.6</td>
<td>1-2 1.05 ± 2.08</td>
</tr>
<tr>
<td>22</td>
<td>41</td>
<td>1.82 ± 1.80</td>
<td>18.2 ± 28.2</td>
<td>1-2 1.22 ± 2.39</td>
</tr>
</tbody>
</table>

1 Mean length of each chromosome expressed as a percentage of the total autosomal length in the cell in which it was identified.

2 Length of short arm divided by total chromosome length \( \times 100 \).

3 Mean total number of autosomal chiasmata per cell of the 41 cells examined: 50.61 ± 3.87 (range: 43-60). For comparison, the mean total number of autosomal chiasmata per cell of 1453 cells from 50 subjects with apparently normal spermatogenesis was 50.07 ± 3.11 (data provided by Drs. A. Chandley, M. Ferguson-Smith, M. Hultén, B. Page, and N.E. Skakkebaek).
This is equivalent to the mean number of chiasmata for each bivalent expressed as a percentage of the total chiasmata number of the cell in which it was identified.

The length of the region of the secondary constriction is subtracted from the length of the chromosome.

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