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Importanza della Placentazione sul peso alla nascita dei Gemelli

L. Gedda e D. Poggi

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	PZ MZ Zigotismo	DZ MZ Zigotismo

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A proposed standard system of nomenclature of human mitotic chromosomes

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The rapid growth of knowledge of human chromosomes in several laboratories, following advances in technical methods, has given rise to several systems by which the chromosomes are named. This has led to confusion in the literature and so to the need for resolving the differences. Consequently, at the suggestion of Dr. C. E. Ford, a small study group was convened to attempt the formulation of a common system of nomenclature. The meeting was arranged, through the good offices of Dr. T. T. Puck, to be held at Denver, in the University of Colorado, under the auspices of the Medical School. The meeting of this study group was made possible by the support of the American Cancer Society, to whom grateful thanks are due. For practical reasons, it was decided to keep the group as small as possible and to limit it to those human cytologists who had already published karyotypes.* In addition, three counselors were invited to join the group to guide and aid the discussions and, if necessary, to arbitrate. Fortunately, the last office did not prove necessary, and it was possible by mutual agreement to arrive at a common system which has flexibility.

It was agreed that the principles to be observed by the system should be simplicity and freedom, as far as possible, from ambiguity and risks of confusion, especially with other systems of nomenclature in human genetics. It should also be capable of adjustment and expansion to meet the needs of new knowledge of human chromosomes. The system should be agreed to by the greatest possible proportion of cytologists working in the field, but the risk that a minority may be unable to accept the system as a whole should not be allowed to delay adoption by a majority.

It was agreed that the autosomes should be serially numbered, 1 to 22, as nearly as possible in descending order of length, consistent with operational conveniences of identification by other criteria. The sex chromosomes should continue to be referred to as X and Y, rather than by a number, which would be an additional and ultimately, a superfluous appellation.

It was generally agreed that the 22 autosomes can be classified into seven groups, distinction between which can readily be made. Within these groups, further identification of individual chromosomes can in many cases be made relatively easily. Within some groups, especially the group of chromosomes numbered 6-12, including also the X chromosome,

* 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 in several or many cells.

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the distinctions between the chromosomes are very difficult to make by presently available criteria. However, lesser difficulties are encountered in separating chromosomes 6 and the X from the remainder of this group. It is believed that, with very favorable prepations, distinction can be made between most, if not all, chromosomes.

It is proposed that the autosomes first be ordered by placing the seven groups as nearly as possible in descending order of size. Within each group the chromosomes are arranged, for the most part, by size. It was desired specifically to avoid the implication that size relationships have been permanently decided in every instance, but it is hoped that the assignment of numbers will be permanently fixed. In those cases where distinction is at present doubtful, final definition of each chromosome can be left until further knowledge has accrued, though an attempt is made to provide a characterization of each. These principles make it possible to draw up a conspectus of the chromosomes, a table of their quantitative characteristics and a table of the synonyms which authors have already published. These are appended (Tables I, II and III).

In Table II, showing the diagnostic characters of the chromosomes, three parameters are relied upon. These are: 1) The length of each chromosome relative to the total length of a normal, X-containing, haploid set, i. e., the sum of the lengths of the 22 autosomes and of the X chromosome, expressed per thousand; 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 two latter indices are, of course, related algebraically quite simply, but it is thought useful to present both here. In some chromosomes, the additional criterion of the presence of a satellite is available (Table I), but in view of the apparent morphological variation of satellites, they and their connecting strands are excluded in computing the indices.

Table II shows the range of measurements determined by various workers. Some of the variation expresses the uncertainty due to measurement of relatively small objects; but many of the discrepancies between different workers' observations are due to the measurement of chromosomes at different stages of mitosis and to the effect of different methods

Tab. I. Conspectus of human mitotic chromosomes

Group 1-3	Large chromosomes with approximately median centromeres. The three chromosomes are readily distinguished from each other by size and centromere position.
Group 4-5	Large chromosomes with submedian centromeres. The two chromosomes are difficult to distinguish, but chromosome 4 is slightly longer.
Group 6-12	Medium sized chromosomes with submedian centromeres. The X chromosome resembles the longer chromosomes in this group, especially chromosome 6, from which it is difficult to distinguish. This large group is the one which presents major difficulty in identification of individual chromosomes.
Group 13-15	Medium sized chromosomes with nearly terminal centromeres (« acrocentric » chromosomes) Chromosome 13 has a prominent satellite on the short arm. Chromosome 14 has a small satellite on the short arm. No satellite has been detected on chromosome 15.
Group 16-18	Rather short chromosomes with approximately median (in chromosome 16) or submedian centromeres.
Group 19-20	Short chromosomes with approximately median centromeres.
Group 21-22	Very short, acrocentric chromosomes. Chromosome 21 has a satellite on its short arm. The Y Chromosome is similar to these chromosomes.

Tab. II. Quantitative characteristics of the human mitotic chromosomes

All measurements were made from cells of normal individuals, except those made by Fraccaro and Lindsten, which included cases of Turner's Syndrome. The column A is the relative length of each chromosome, B is the arm ratio and C the centromere index, as defined in the text.

	Tjio and Puck (6)			Chu and Giles (2)			Levan and Hsu (5)			Fraccaro and Lindsten*			Lejeune and Turpin (4) *			Buckton, Jacobs and Harnden *			Range		
	A	B	C	A	B	C	A	B	C	A	B	C	A	B	C	A	B	C	A	B	C
1	90	1.1	48	90	1.1	48	85	1.1	49	82	1.1	48	87	1.1	48	83	1.1	48	82-90	1.1	48-49
2	82	1.6	39	83	1.5	40	79	1.6	38	77	1.5	40	84	1.5	40	79	1.6	38	77-84	1.5-1.6	38-40
3	70	1.2	45	72	1.2	46	69	1.2	45	65	1.2	45	67	1.2	46	63	1.2	46	63-72	1.2	45-46
4	64	2.9	26	63	2.9	26	63	2.7	27	62	2.6	28	62	2.6	25	60	2.6	28	60-64	2.6-2.9	25-28
5	58	3.2	24	58	3.2	24	59	2.6	28	60	2.4	29	57	2.4	30	57	2.4	30	57-60	2.4-3.2	24-30
X	59	1.9	34	57	2.8	38	52	1.6	38	54	1.6	38	58	2.2	32	51	1.7	37	51-59	1.6-2.8	32-38
6	55	1.7	37	56	1.8	36	56	1.7	37	54	1.6	38	56	1.7	37	56	1.6	38	54-56	1.6-1.8	36-38
7	47	1.3	43	52	1.9	35	51	1.9	35	50	1.7	37	51	1.8	36	50	1.7	37	47-52	1.3-1.9	35-43
8	44	1.5	29	46	1.7	29	48	1.6	33	47	1.7	37	48	2.4	29	46	1.5	40	44-48	1.5-2.4	29-40
9	44	1.9	40	46	2.4	38	47	1.8	36	45	2.0	33	47	1.9	35	44	2.1	32	44-47	1.8-2.4	32-40
10	43	2.4	27	45	2.3	30	45	2.0	33	45	2.6	34	45	2.6	27	44	1.9	35	43-45	1.9-2.6	27-35
11	43	2.8	34	44	2.1	32	44	2.2	31	43	2.2	31	44	1.6	39	43	1.5	40	43-44	1.5-2.8	31-40
12	42	3.1	24	43	3.1	24	42	1.7	32	43	1.7	37	42	2.8	27	42	2.1	32	42-43	1.7-3.1	24-37
13	35	8.0	11	32	9.7	10	32	5.0	16	34	4.8	17	33	6.8	14	36	4.9	17	32-36	4.8-9.7	10-17
14	32	7.3	12	34	9.5	9	37	4.0	18	35	4.4	19	32	7.0	13	34	4.3	19	32-37	4.3-9.5	9-19
15	29	10.5	9	31	11.9	8	35	4.7	17	33	4.6	22	31	10.0	9	34	3.8	22	29-35	3.8-11.9	8-22
16	32	1.8	36	27	1.6	38	30	1.4	42	31	1.4	42	29	1.4	41	33	1.4	31	27-33	1.4-1.8	31-42
17	29	2.8	26	30	2.1	33	29	2.4	30	30	1.9	35	29	3.1	23	30	1.8	36	29-30	1.8-3.1	23-36
18	24	3.8	21	25	3.8	22	25	2.6	28	27	2.5	29	26	4.2	21	27	2.4	29	24-27	2.4-4.2	21-29
19	22	1.4	41	22	1.9	34	24	1.2	40	25	1.3	43	22	1.4	42	26	1.2	45	22-26	1.2-1.9	34-45
20	21	1.3	44	19	1.3	44	21	1.2	40	23	1.3	43	20	1.2	43	25	1.2	46	19-25	1.2-1.3	40-46
21	18	3.7	21	15	6.8	13	13	2.5	28	19	2.5	29	15	2.3	31	20	2.5	29	13-20	2.3-6.8	13-31
22	17	3.3	23	12	6.0	14	16	2.0	33	17	2.3	30	13	4.0	20	18	2.7	27	12-18	2.0-6.0	14-33
Y	19	—	0	11	—	0	18	4.9	17	22	2.9	26	18	—	0	18	4.9	17	11-22	2.9- —	0-26

* Unpublished data.

Tab. III. Synonymy of chromosomes as published by various workers

New Chromosome Number	Tjio and Puck (6)	Chu and Giles (2)	Levan and Hsu (5)	Ford Jacobs and Lajtha (3)	Böök Fraccaro and Lindsten (1)	Lejeune, Turpin and Gautier (4)
1	1	1	1	1	1	G1
2	2	2	2	2	2	G2
3	3	3	3	3	3	G3
4	4	4	4	4	4	G4
5	5	5	5	5	5	G5
6	6	6	6	6*	6	M1
7	7	7	7	(8)	7	M2
8	8	8	8	(9)	8	Md1
9	9	9	9	(11)	9	M3
10	10	10	10	10	10	Md2
11	11	11	11	(12)	11	M4
12	12	12	12	(13)	12	Md3
13	18	14	20	14	14	T1
14	19	15	18	15	15	T2
15	20	13	19	16	13	T3
16	13	17	15	19	16	C1
17	14	16	13	17	17	P1
18	15	18	14	18	18	P2
19	16	19	16	20	19	C2
20	17	20	17	20	20	C3
21	21	21	22	22	21	Vh
22	22	22	21	23	22	Vs
X	X	X	X	? (7)	X	X
Y	Y	Y	Y	Y	Y	Y

* In the published idiogram the chromosomes of group 6-12 (including X) were indicated by discontinuous lines and left unnumbered owing to the uncertainty of discrimination at that time. For the purpose of this table, these chromosomes have been assigned the numbers shown in brackets, in serial order of length.

of pretreatment and preparation for microscopic study. The ranges shown, therefore, represent the maxima and minima of the means found by different workers using different techniques. However, within any one worker's observations, the variations are not so broad.

Reference should be made to two other matters of nomenclature. In the first place, it is considered that no separate nomenclature for the groups is needed. It is considered that any group to which it may be necessary to refer will be a sequence of those designated by Arabic numerals. Hence, any chromosome group may be referred to by the Arabic numerals of the extreme chromosomes of the group, joined together by a hyphen, e. g., the group of the three longest chromosomes would be Group 1-3. This scheme has the merit of great

flexibility. For instance, chromosomes X and 6 may be separated from the Group 6-12 whenever they can be distinguished.

Secondly, there is the problem raised by the abnormal chromosomes which are being encountered in the more recent studies. Their nomenclature was discussed wothout a definite conclusion being reached. Broadly, it was agreed, however, that any symbol used should avoid incorporating a specific interpretation which was not reasonably established. It was suggested that arbitrary symbols, prefixed by a designation of the laboratory of origin, should usually be assigned to the abnormal chromosome.

In this connection, two further requisites for coordination of research were discussed. One is the storage of documentation for reference, perhaps in a central depository, additional to what it may be possible to publish. The other is the desirability that cultures be preserved, by the satisfactory methods now used, so that they are available for reference, comparison and exchange.

Some consideration was also given to the desirability of using a uniform system for presenting karyotypes and idiograms, but recognizing that individual variation in taste is involved, rigidity of design was thought undesirable. However, it was recommended that the chromosomes should be arranged in numerical order, with the sex chromosomes near to but separated from the autosomes they resemble. It is desirable that similar ones be grouped together with their centromeres aligned.

It is recognized that choice between the different possible schemes of nomenclature is arbitrary, but that uniformity for ease of reference is essential. Hence, individual preferences have been subordinated to the common good in reaching this agreement. This human chromosomes study group therefore agrees to use this notation and recommends that any who prefer to use any other scheme should, at the same time, also refer to the Standard System proposed here.

We are well aware of the wide interest in the work of this study group and realize that this meeting is merely a preliminary to a larger meeting. It is believed that two needs have to be met in this respect. One is for seminars and workshops at which workers in the field may exchange information; such seminars are best arranged regionally. The second need, which may come later, is for international conferences; and we believe that Congresses and other organizations whose interests include human genetics, should promote such meetings.

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