

## PREFACE

I was fortunate to have had the idea that led to this book at the right time and at the right place! During the first half of 1974, while I was on the faculty of the Johns Hopkins University School of Medicine in Baltimore, I was a Visiting Fellow at Yale University in New Haven. I was discussing the use of chromosomally abnormal human cell lines for regional assignment of loci in somatic cell hybridization (see, e.g., Borgaonkar et al, 1973; Borgaonkar et al, 1977; Denney, Borgaonkar, and Ruddle, 1978), and questions arose about what material was available and how that information should be stored. A systematic compilation of chromosomally abnormal material was necessary.

The idea to computerize information on abnormal karyotypes in an organized fashion, which could then be retrieved to compile a Catalog, was conceived during the discussions I held with my then colleagues at both Yale and Johns Hopkins [Borgaonkar, Bolling, Partridge, Ruddle, and McKusick, 1975]. Significant improvements and changes have been made since then, partly because of my discussions with colleagues during the last 23 years.

I believe the Atma (A Sanskrit word meaning, roughly, the immortal soul or spirit of an individual person, place, or thing) of the Catalog is in the methodology of organization, its simplicity of use, and the ease and economy with which it can be updated. The speed with which these eight editions have been prepared provides not only gratifying proof that the Catalog embodies these characteristics but also demonstrates that up-to-date editions can be and are promptly prepared. In addition, on-line access to current data has been provided since 1980, first through the computing facilities of the University of Delaware. Since 1994, the entire data base has been on a laptop as well as on-line through the facilities of the Medical Center of Delaware. Plans are being made to make this data base available on the Internet.

The first section of the Catalog, which is necessarily a major component because of the amount and nature of the material available, is concerned with structural chromosomal variations and anomalies such as deletions, inversions, and translocations. Entries in the Catalog are listed according to the chromosome break points. The first two columns of the entry refer to the chromosome (01 to 22, 0X and 0Y); the third column refers to the chromosome arms (p and q); and the fourth and fifth columns of the entry number refer to the region and band, respectively. Whenever information corresponding to the latter three columns is not available, a 0 is entered in the appropriate column. If and when further information on such an entry becomes available, the revised information can easily be substituted. Following the International System for Human Cytogenetic Nomenclature [ISCN, 1995] recommendations whenever a band is subdivided into units, I have entered the information accordingly in the sixth and seventh columns; e.g., 06p2105 implies a break halfway in the band 06p21. Alternatively, if information is available on sub-bands it is entered in the sixth column; if not, a 0 is entered. Thus, the first autosome item encoded is 010000 and the last is 22q1333. In other words, the minimal information required for a report to be included in this section of the Catalog is that the chromosome involved in the production of the variation or anomaly be known. If an entry is cited under a specific band, generally it is because the authors of the report used at least one of the banding techniques to enable them to designate the break point. Under each category all the reports are listed alphabetically according to the

## Preface

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last name of the first author. The bibliographic citation is followed by chromosome constitution(s) of the individual(s) reported. When available, to avoid possible confusion arising from multiple reporting of the same case, the subjects are identified by their case numbers. I would urge those who collate data to give due consideration to this matter. Some rare translocations and inversions, when reported without citation to an earlier published report, have created an erroneous impression that there are many such cases and therefore, break-points at those chromosome sites.

In general, a policy of listing most of the recent reports of structural aberrations has been adopted. References prior to 1980 can be found in the bibliographies of the more recent reports cited in this Catalog. However, certain references prior to this date have been maintained in this edition of the Catalog on the basis of their uniqueness, priority, etc.

The entries on chromosomes are arranged in numerical order. Whenever a reference has information on two or more break points (or abnormalities), an appropriate comment is inserted at the second point of entry. For example, the translocation  $t(5;14)(p14;q21)$  will have its complete entry at 05p140 (Borgaonkar et al, 1973) and at 14q210 its secondary entry will have the following notation: "Same entry as in 05p140 (Borgaonkar et al, 1973)." If the break points were not identified precisely—e.g., in reports of ring chromosomes and pericentric inversions—then such information has been entered only for the p area of the relevant chromosome. There is no secondary entry for such reports on the q side of the same chromosome.

The second section of the Catalog lists numerical anomalies including trisomies, monosomies, and polyploids. The first two digits of the entry refer to the chromosome number, and the third column has either a plus (+) or a minus (–) sign, indicating trisomy or monosomy, respectively. References on polyploidy are arranged in alphabetical order under triploidy and tetraploidy.

The third section of the Catalog includes comments on conditions that are termed "Chromosomal Breakage Syndromes." Since the aberrations are nonspecific for a single chromosome, they have been entered under a separate category and are listed in alphabetical order.

One of the uses of the Catalog is in chromosome mapping and gene assignment. Therefore, it is desirable to include information on the availability of chromosomally mutant cell lines because of their use in somatic cell hybridization studies. This was made possible by my fortunate collaboration, in 1974, with Drs. Coriell and Greene of the Coriell Institute for Medical Research, Camden, New Jersey. Included in the Catalog are the listing of chromosomally abnormal cell lines (along with their identifying numbers) available from their Repository as well as some other sources whenever those data were available to me.

In compiling the Catalog, some arbitrary decision making became necessary. Some of these decisions may have to be reconsidered as additional information becomes available.

- 1) In part because of the use of the various banding techniques, more and more structural aberrations are being detected. However, if no specific break points were described in the report but could be deciphered easily from the figures or from the content of the paper, then the inferred break points were cataloged. Figure 1 shows that all of the 86 chromosome regions (as defined by ISCN [1995]) were involved in interchanges [also see Kamat and Borgaonkar, 1979]. The importance

of position effect phenomenon in individuals with balanced translocations with phenotypic consequences can be evaluated by analyzing such data in more detail [Borgaonkar, 1973].

It is important to note that two microscopically similar translocations—identical as per ISCN [1995]—may in fact be genetically different, since the break points may be a few loci apart. Obviously, a similar situation holds for other types of structural anomalies. Because one of the uses of the Catalog is in gene mapping, and because more reports of structural anomalies are to be found, the structural anomaly section of the Catalog is much more detailed than other sections. However, by no means all reports are cited in the Catalog (for example, reports on ring chromosome 18). As elsewhere in the Catalog, it has been the goal to provide key references on all kinds of chromosomal variants and anomalies, and through these, readers should be able to locate the literature on any given condition or topic. Since the data base is on-line we can do a search on any topic including translocations, rings, and deletions etc., and obtain the information. Recently, however, newer techniques such as spectral karyotyping of chromosomes have been developed and some previously undetected but suspected anomalies were confirmed to be minor(?) structural rearrangements.

- 2) Of the numerous reports on relatively common conditions such as the trisomies of chromosomes 13, 18, and 21, partial monosomies of 5p and 18p, and the 45,X and 47,XXY chromosomal abnormalities, only a few reports have been cataloged on the basis of priority, uniqueness, or general coverage.
- 3) Reports on chromosome changes in tumors, cancers, and leukemic cell lines have been almost entirely excluded.
- 4) Data on experimentally reproduced chromosomal break points have also been excluded.

Regarding the use of nomenclature, I have attempted to adhere to the guidelines put forward by the ISCN [1995] and its predecessors. Several departures or modifications seem appropriate, however.

- 1) In the aneuploidy section, the first two digits in the entry for the chromosome number have been maintained. These are followed by the designations (+) for a trisomy and (-) for a monosomy. Double aneuploidy is entered in the same manner. In this section all entries are in numerical order, followed by X and Y. For multiple sex chromosomal anomalies, those of the X chromosome are listed first.
- 2) In translocations where only changes in length were known, single parentheses are used—e.g. 46,XX,t(5p+;13q-). No assumption is made with regard to the reciprocal nature of a translocation unless this was so stated in the report. A statement of balanced translocation in a report was not interpreted as necessarily implying a reciprocal translocation!
- 3) Whenever an existing band is subdivided, the ISCN [1995] recommendations call for a decimal point to be placed after the original band designation, followed by the number assigned to the sub-band. The decimal point has not been used in the entry numbers. There have been changes in the band and sub-band designations of certain areas during the evolution of ISCN in the last two decades. I entered the data

Preface

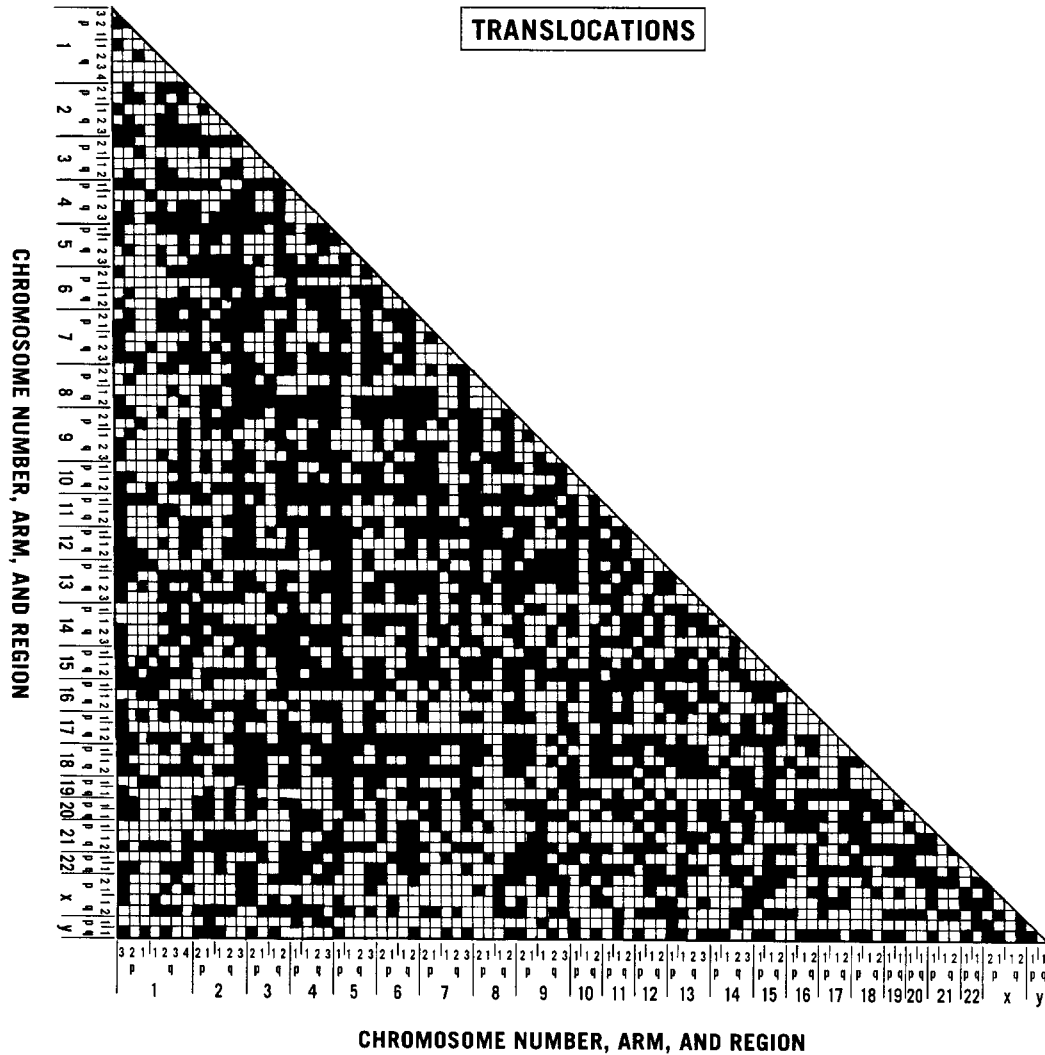


Fig. 1. Chromosomal translocations between different segments. (Prepared by Dr. Cara Gatto-Weiss, Christiana Care Health System.)

on citations when those report came to my attention. Therefore, certain band designations are not going to be along the lines of the present ISCN, i.e., 1995. Earlier reports using the then current ISCN versions have used the band designations that existed at that time e.g., 3p27. The band 3p27 does not exist in the 1995 ISCN edition. I have chosen to keep those entries intact in the data base for various reasons. Reinterpretations of the breakpoint data of these earlier entries in the data base will lead to further confusion. It may be possible to edit such entries in future and make appropriate changes when and if more accurate information on breakpoints becomes available in these cases.

To facilitate the collection of data regarding preferential involvement of certain chromosomal regions in aberrations, the entry number has been included with the banding pattern for the band or sub-band—i.e., Negative, Positive, or Variable as per the Q and G banding techniques. As we begin to understand the biochemical basis of the banding techniques and its relationship to chromosome breakage, this information may be helpful in understanding the mechanics of aberration (Table I). Admittedly, the data in Table I reflect only those break points that are reported in the literature and entered in the Catalog. Considerable bias has gone into this compilation. Even then, the difference between the expected involvement of the three band regions (Negative, Positive, and Variable) based on their proportionate occurrence in the genome and that actually observed in the breakpoints has been demonstrated to be statistically significant [Yu et al, 1978; Porforio et al, 1987].

In addition to the reasons stated earlier [Borgaonkar and Bolling, 1976 when the data base was being established, and more recently by me [Borgaonkar, 1994], there were other significant reasons for undertaking a computerized chromosomal Catalog.

- 1) The geneticist may pool linkage data obtained by the pedigree approach using marker chromosomes (Tables II and VI). The Catalog could be used in the selection of cell lines for regional assignment of loci by the somatic cell hybridization approach.
- 2) In the description of new clinical material with chromosomal abnormalities, it is becoming increasingly difficult to ascertain from the literature what, if anything, has been published on the same type of chromosomal abnormality. The information that is available in journals is not easily culled. For example, a father of a young girl wrote to us from Norway during the time the Winter Olympics were being held there in 1993, asking for information on citations of ring chromosome 15. An examination of the data in Table VI shows that there are at least 32 reports of this anomaly in this data base. It is hoped that the Catalog will satisfy such needs. I have included a list of chromosome band numbers (Table II) that are known to be polymorphic by the nature of their staining properties with various banding techniques such as quinacrine fluorescence, constitutive C-banding heterochromatin,

**TABLE I. Number of Entries in the Catalog by Type of Band and Break Points**

Edition	Negative Band n (%)	Positive Band n (%)	Variable Band n (%)	Total
1st — 1975	450 (64.4)	101 (14.4)	148 (21.2)	699
2nd — 1977	907 (65.5)	264 (19.1)	214 (15.4)	1385
3rd — 1980	1554 (67.63)	487 (21.29)	257 (11.18)	2298
4th — 1983	2343 (67.04)	765 (21.89)	387 (11.07)	3495
5th — 1988	4034 (65.04)	1613 (26.01)	555 (8.95)	6202
6th — 1991	5097 (66.73)	1929 (25.26)	612 (8.01)	7638
7th — 1993	6091 (68.00)	2179 (24.32)	688 (7.68)	8958
8th — 1997	8028 (66.28)	2762 (22.8)	1322 (10.91)	12,112
Proportion in genome	54.2	38.2	7.6	

## Preface

**TABLE II. Polymorphic (or Variant) and Fragile Site Areas in the Human Genome**

Polymorphic:					
1q120	4p120	9q120	15p100	17p120	22p130
1q210	4p163	9q130	15p120	17p130	22q110
2q110	4q350	9q211	15q110	17p133	Xp2232
2q120	5q112	9q320	15q112	18p110	Xq130
2q130	6p110	11p110	15q120	19q100	Xq280
2q370	6q110	11q232	16p112	20p130	Yp110
3p110	6q150	13p100	16q000	20q110	Yq110
3p140	6q270	13p110	16q110	21p100	Yq112
3q110	9p120	13p130	16q112	21p130	Yq120
4p110	9p130	14p100	16q221	22p100	
Fragile Site Areas:					
1p110	3p140	7q220	11p130	16p120	Xp220
1p213	3p142	7q310	11p151	16p130	Xp222
1p310	3p210	7q311	11q130	16q210	Xq220
1p360	3q262	7q320	11q230	16q220	Xq260
1q250	4q340	8q220	11q231	16q221	Xq263
1q410	5q130	8q241	11q233	16q230	Xq270
1q420	5q310	9p210	12q130	16q240	Xq271
2p240	5q350	9q310	12q2413	17p120	Xq272
2q110	6p230	9q320	12q242	17q210	Xq273
2q112	6q260	10q230	13q310	19p130	Xq280
2q130	7p130	10q241	14q230	20p110	Yq120
2q310	7p220	10q250	15q220	22q130	

and DAPI. Also included in this table are fragile sites that have been elicited by special media in tissue culture. Availability of such lists has been found to be particularly useful in a variety of situations, such as occasions when a variant pattern is found in a case and the investigator becomes aware that family studies may be warranted [Borgaonkar, 1995, 1996].

The Catalog is useful in collation of information while describing new chromosomal syndromes. In 1975, we [Borgaonkar et al, 1976] had considered the possibility of a ring chromosome 20 syndrome on the basis of three patients with behavioral problems, mental retardation, and EEG changes. Subsequently, Lancman et al [1993]; Holopainen et al [1994] and van Langen et al [1996] have collated data on several more patients with r(20) and have confirmed our initial description of this condition. Reports assembled in the Catalog are so arranged that monosomic and trisomic reports of chromosomes or of chromosomal regions can be located systematically (Table VI). Partial chromosomal aneuploidy syndromes are tabulated in Table III and aneuploidies involving whole chromosomes are listed in Table IV. Collation of reports on chromosomal variants, anomalies, and aneuploidies has been found to be useful and is of considerable value in genetic counseling of patients and families with chromosomal problems [Borgaonkar, 1996]; knowledge about the phenotype of carriers of seemingly similar balanced translo-

**TABLE III. Chromosome Arms Involved in Partial Aneuploidy Syndrome (- Absent; + Present)**

Arm	Monosomy	Trisomy	Arm	Monosomy	Trisomy
1p	+	+	11p	+	+
1q	+	+	11q	+	+
2p	+	+	12p	+	+
2q	+	+	12q	+	+
3p	+	+	13q	+	+
3q	+	+	14q	+	+
4p	+	+	15q	+	+
4q	+	+	16p	+	+
5p	+	+	17p	+	+
5q	+	+	17q	+	+
6p	+	+	18p	+	+
6q	+	+	18q	+	+
7p	+	+	19q	-	+
7q	+	+	20p	+	+
8p	+	+	21q	+	+
8q	+	+	22q	+	+
9p	+	+	Xp	+	+
9q	+	+	Xq	+	+
10p	+	+	Yp	+	+
10q	+	+	Yq	+	+

cation or inversion carriers has been an important factor in parents' decisions to continue a pregnancy.

It appears that we humans as a biological species are better able to "tolerate" extra chromosomal material than the lack of it (Table IV). Thus, fewer monosomies than trisomies have been documented so far in chromosome studies on abortuses, stillborns, and liveborns. This impression of ours has since been confirmed by data reported in this book in the last decade.

- 3) In structural aberrations it will be possible to relate the involvement of the type of band (i.e., Negative, Positive, or Variable) to the occurrence of a certain type of aberration in the human karyotype; in other words, there will be an extension of earlier studies [Yu et al, 1978].
- 4) The chromosomal variations and anomalies were placed into several categories, which were then coded for computerization and retrieval purposes (Tables V and VI). The nomenclature of the categorization mostly follows that of the ISCN [1995] report. Into which group an aberration falls is determined by examining the chromosome constitution of the individuals reported. This information has been retrieved from the data base and tabulated. Table VI includes, for each chromosome, the number of aberrations listed. This should facilitate a search for, and compilation of, data on the various types of abnormalities that occur in chromosomes; for

## Preface

**TABLE IV. Chromosomes Involved in Full Aneuploidies**

Chromosome	Monosomy	Trisomy
1	+	+
2		+
3		+
4		+
5		+
6		+
7		+
8		+
9		+
10		+
11		+
12		+
13	+	+
14	+	+
15	+	+
16	+	+
17		+
18	+	+
19		+
20	+	+
21	+	+
22	+	+
X	+	+
Y	+	+

example, an examination of Table VI demonstrates that ring chromosome has been described seven times for chromosome 16.

- 5) The assemblage of data on abnormal karyotypes in this Catalog, makes it possible to work with problems related to the estimation of risk figures in the transmission of abnormal rearrangements and involvement of factors such as sex and bias of ascertainment. Khaldi et al [1979] presented risk estimates for inversion carriers, and Petrosky [1983] and Petrosky and Borgaonkar [1984] for translocation carriers.
- 6) It was my intention to establish an International Registry of Abnormal Karyotypes according to the method of this Catalog. Accordingly, the Fourteenth Listing of the Repository of Human Chromosomal Variants and Anomalies was published in 1993 [Borgaonkar et al, 1993].

The Repository consists of personally communicated karyotypic findings, whereas the Catalog is a selected listing, with comments and interpretations, of published reports on all significant kinds of chromosomal variants and anomalies. The Repository was intended to be a record of all the findings (including published papers and unpublished studies) on chromosomally normal and abnormal individuals of those laboratories reporting, voluntarily, their data to us. The data in the Repository are coded for techniques used, mode of ascertainment, kind of anomaly, and the locale of the reporting laboratory, whereas the Catalog is

**TABLE V. Aberration Codes\***

IC	Isochromosome The two arms of the chromosome are identical to each other.
TD	Terminal deletion A terminal segment of a chromosome is deleted.
ID	Interstitial deletion An intermediary segment, i.e., excluding a centromere and terminal ends (telomeres), of a chromosome is deleted.
IP	Inversion paracentric An inversion of a chromosome segment that excludes the centromere.
PI	Inversion pericentric An inversion of a chromosome segment that includes the centromere.
RI	Ring chromosome Two broken ends of a chromosome have joined to form a ring-like structure.
DI	Dicentric chromosome A chromosome with two centromeres.
RT	Reciprocal translocation In a translocation, the segments of chromosomes have been exchanged.
TR	Robertsonian translocations Translocations involving the centromeric regions and with both long arms of acrocentric chromosomes.
IN	Direct insertions within a chromosome A segment has been inserted into the same chromosome at another point.
II	Inverted insertions within a chromosome An inverted segment has been inserted into the chromosome at another point.
IX	Direct insertion between two chromosome A segment from one chromosome has been inserted into another chromosome.
XI	Inverted insertions between chromosomes An inverted segment from one chromosome has been inserted at a point into another chromosome.
CT	Complex translocation A translocation involving three or more breakpoints between two or more chromosomes.
ST	Simple translocation A transfer of a segment between two chromosomes.
DU	Duplication A chromosome in which a segment is present in duplicate.
TX	Tandem translocation A transfer of terminal segments between chromosomes.
RE	Recombinant chromosomes A structurally rearranged chromosome with a new segmental composition resulting from meiotic crossing-over.
MA	Marker chromosome This term includes chromosomes with polymorphisms and variant features.
WT	Whole-arm translocations Whole-arm exchanges involving nonacrocentric chromosomes.
TA	Terminal rearrangements Two chromosomes are joined end-to-end.
DD	Direct duplication A duplicated segment is found on the same chromosome.
CF	Centromeric fission Splitting of the centromere resulting in two functional centromeres.
DT	Double translocation Two independent translocations in the same individual.

*(continued)*

## Preface

**TABLE V. Aberration Codes** (*continued*)

UT	Unstable translocation When translocations are constantly changing.
FS	Fragile sites A fragile site elicited by any of the techniques in the genome.
DA	Double aberration When two or more anomalies are present in one individual.
UP	Uniparental disomy A condition in which both homologous chromosomes are from one parent.
SA	Satellited chromosome A satellited area on a chromosome that is not a short arm of acrocentric chromosomes 13, 14, 15, 21, and 22. (These five areas are not coded in this Catalog as they are well-established characteristics of these five chromosomes.)
IS	Isodisomy A condition in which both the chromosomes are duplicates of one another within a diploid genome.

\*For detailed explanations of these codes the reader could consult the ISCN (1995) and other standard texts in cytogenetics.

coded for kind of anomaly, chromosome aneuploidy, and involvement of Mendelian conditions. Plans are underway to make the Repository/Registry data base available on the Internet and anyone interested in this matter should contact me and/or look for announcements.

This Eighth Edition of the Catalog is available from the Wiley-Liss Division of John Wiley & Sons, Inc., 605 Third Avenue, New York, New York 10158-0012. Plans are underway to make the Catalog database available on Internet. Anyone interested in this matter can contact me or John Wiley & Sons, Inc. and look for announcements for further details. For further information please contact Dr. Digamber S. Borgaonkar, Cytogenetics Laboratory, P.O. Box 6001, Christiana Care Health System, Newark, DE 19718, USA; Fax 302-733-3773; Telephone 302-733-3530; e-mail internet:borgaonkar.d@mcd.gen.de.us

A Catalog such as this probably would not have been meaningful prior to the significant advances made in karyotyping techniques. The banding techniques of the 1970's enabled us to know more about the nature and extent of variations and anomalies in chromosome segments. Techniques such as FISH and PRINS have allowed us to detect microdeletions and submicroscopic rearrangements (e.g., Table VII). These types of studies have been used in gene mapping. Note the growth of data in Tables VIII and IX citing these data. The incidence of chromosomal variations and anomalies in liveborns seems to be in the vicinity of 5%, and in zygotes it is several times greater than this figure. Prior to the advent of banding techniques, it was thought that there were several chromosomes (e.g., 19 and 20) "spared" from involvement in aberrations, partly because of the nature of genetic material present on them. I have shown elsewhere that each and every chromosome has been implicated in interchanges [Borgaonkar, 1975]. In the second edition of the Catalog a similar study showed that all 48 chromosome arms were involved in interchanges. In 1979 we showed that each and every region in the human genome is known to be involved in producing variation [Kamat and Borgaonkar, 1979]. Almost all the bands [ISCN, 1995] are involved in chromosomal interchanges. In the past, several cytogeneticists have attempted to collate data with a view to facilitate the karyotype-phenotype relationships in this discipline. Some had collected this material and presented it in a concise form. For example,

**TABLE VI. Numbers of Different Aberrations for Various Human Chromosomes**

Aberration Code	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	X	Y	Total
IC	1	1	0	1	5	1	2	7	24	2	0	48	4	7	14	0	3	54	0	11	13	4	66	27	295
TD	30	22	27	87	41	19	33	27	46	40	41	30	22	27	87	41	19	33	27	46	40	41	88	43	957
ID	43	46	22	46	34	36	58	40	20	20	55	9	65	14	92	19	46	13	1	9	12	56	56	6	818
IP	23	12	21	6	22	23	31	14	9	11	18	10	7	19	8	3	8	3	2	1	2	1	13	0	267
PI	46	59	39	38	47	28	22	29	42	32	18	15	25	6	12	12	9	18	10	10	8	8	22	22	577
RI	9	6	10	24	8	17	11	6	21	15	7	7	48	25	32	7	16	29	10	22	39	30	31	25	455
DI	1	0	0	0	4	1	1	3	13	1	0	1	16	6	23	1	1	11	0	2	10	13	56	54	218
RT	271	239	207	229	173	137	150	97	131	100	112	58	61	47	47	22	22	24	9	7	3	1	86	26	2259
TR	0	0	0	0	0	0	0	0	0	0	0	0	98	48	33	0	0	0	0	0	50	9	0	1	239
IN	3	2	4	2	3	0	2	2	2	1	1	0	1	0	1	0	0	1	0	0	0	0	2	0	27
II	3	6	4	1	5	1	1	9	4	0	1	2	2	0	0	1	1	1	0	0	0	0	0	1	43
IX	9	9	10	3	8	4	6	2	2	3	4	3	3	3	2	0	0	0	0	1	1	0	0	0	73
XI	4	3	7	3	2	0	1	1	0	0	0	1	0	0	0	0	0	0	1	0	0	0	0	0	23
CT	23	25	12	6	7	7	8	2	5	0	4	1	1	1	1	0	0	0	0	1	1	3	1	1	109
ST	146	118	117	133	104	91	79	57	109	53	71	32	53	35	44	14	17	30	8	10	10	5	192	72	1600
DU	10	16	5	9	7	5	13	23	18	10	2	8	5	8	24	5	15	8	0	0	8	9	28	1	237
TX	1	0	0	0	0	1	0	0	0	0	1	1	3	1	2	0	0	0	0	0	12	0	7	1	30
RE	2	1	5	7	8	2	3	8	0	5	1	0	6	1	0	2	1	4	0	1	2	4	1	0	64
MA	22	9	9	7	4	6	1	1	36	1	3	2	11	8	25	19	17	7	3	4	17	14	5	51	282
WT	18	7	2	7	3	6	4	2	8	2	1	5	6	4	5	0	0	7	0	0	1	0	3	2	93
TA	1	0	0	0	0	0	0	0	1	1	0	0	1	0	0	0	0	1	0	0	2	0	4	2	13
DD	8	15	2	4	0	0	7	5	3	3	5	4	0	2	1	2	5	3	1	0	1	1	5	0	77
CF	0	0	0	3	1	0	4	1	0	1	0	0	1	0	0	0	0	0	0	2	0	0	0	0	13
FS	10	12	11	2	4	4	6	4	5	16	10	10	1	1	1	26	7	0	3	1	0	2	78	1	215
DT	3	3	4	2	2	1	3	0	0	0	1	0	1	0	0	0	0	0	0	0	0	0	0	0	20
UT	0	0	0	0	1	0	0	0	0	0	1	0	0	0	1	0	0	0	0	0	0	0	0	0	3
DA	0	0	0	0	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0	1
UP	0	0	0	0	0	0	2	1	0	1	2	0	2	5	14	3	0	0	0	0	2	2	2	0	36
SA	0	1	0	3	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	2	7
IS	0	1	0	0	0	0	2	0	0	0	2	0	1	1	1	1	0	0	0	0	0	0	0	0	9
Total	687	613	518	623	493	391	450	341	499	318	361	247	445	269	470	178	187	247	75	125	236	201	748	338	9060

**TABLE VII. Reports of Structural Aberrations, Polymorphisms, and Fragile Sites in Successive Editions of the Catalog**

	TOTAL	IP*	ID†
First Edition - 1975	862	1	5
Second Edition - 1977	1,480	2	27
Third Edition - 1980	2,082	5	50
Fourth Edition - 1984	3,041	23	123
Fifth Edition - 1989	4,871	99	330
Sixth Edition - 1991	6,522	149	487
Seventh Edition - 1993	7,596	170	618
Eighth Edition - 1997	9,080	267	818

\*IP = paracentric inversion.

†ID = interstitial deletion.

## Preface

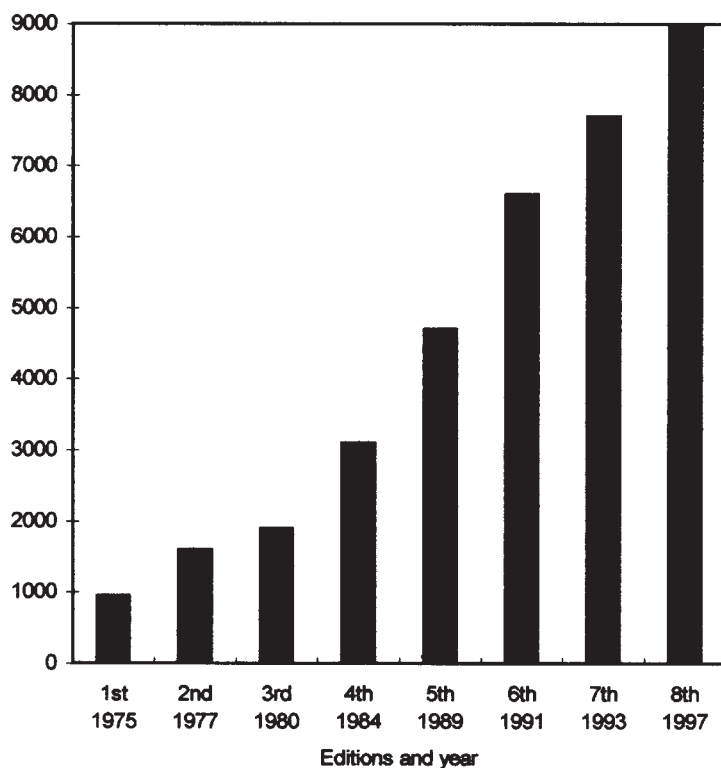


Fig. 2. Growth of data on structural chromosome aberrations, polymorphisms, and fragile sites as seen in editions of this catalog.

Thompson [1965, 1966] compiled the data on autosomes into a catalog; Hamerton [1971] and Baserga et al [1973] tabulated large amounts of data on abnormal karyotypes in their respective books; Yunis [1977] edited *New Chromosomal Syndromes*. DeGrouchy and Turleau [1977] prepared the *Clinical Atlas of Human Chromosomes*, and have published a second edition [1982]. Schinzel [1983] cataloged effects of unbalanced chromosome aberrations in man. In the Birth Defects Encyclopedia [Buyse, 1990] there were detailed descriptions of chromosomal syndromes. Because of the advances in techniques for studying chromosomes, however, the literature is inundated with reports of all types of anomalies. Some specialty journals that publish individual case reports with chromosomal anomalies have somewhat reluctantly encouraged authors to collate data on similarly affected patients. Thus, a survey was frequently out of date by the time it was published. A more convenient and efficient system for retrieving information on abnormal karyotypes was necessary, and we believe that this data base fulfills this need. In the last few years, several Mendelianizing conditions have been known to be chromosomal aneuploidy syndromes as well. Examples are DMD (Duchenne Muscular Dystrophy) and interstitial deletion of a segment of band Xp21; Miller Dieker syndrome and band 17p13; retinoblastoma and band 13q14. [See citation of

Lele et al (1963) under 13q000 in this book, which is an exception and excellent example of astute observation.] Initial work led to suspicious localization of the gene to these segments and fine tooling led to sequencing the gene, e.g., RB1. Because of such developments [Collins, 1995] and their potential use, we coded the reports of chromosomal studies in Mendelian disorders with the MIM numbers and tables have been prepared listing these reports (Tables VIII and IX). The computerized human chromosome data bank is only one step toward meeting the problem of literature searching. It has become a tool for other studies as well (e.g. Porfirio et al, 1987). Keeping up with the data being published and cataloging it in this data base has been a constant activity for more than two decades. The availability of the data base on-line for others to tap into will be a dream come true.

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## Preface

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