湖南医学院学报 专辑

中国人类染色体异常目录

（一）

A CATALOGUE OF HUMAN CHROMOSOMAL ANOMALIES IN CHINA—I

1986·4

A Catalogue of Human Chromosomal Anomalies in China

中国人类染色体异常目录

Ist Collection

第1集

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Contents:

目 录

Preface
前言 ……(1)
Guide to the use
使用说明 ……(5)
Registry of chromosomal examination
染色体检查登记 ……(7)
Catalogue of chromosomal anomalies
染色体异常目录 ……(21)
Chromosome
染色体
1……(23)
2……(25)
3……(27)
4……(30)
5……(34)
6……(37)
7……(40)
8……(42)
9……(43)
10……(45)
11……(47)
12……(48)
13……(49)
14……(51)
15……(52)
16……(53)
17……(54)
18……(54)
19……(54)
20……(55)
21……(55)
22……(56)
X ……(56)
Y ……(58)
Appendix: Author index
附录： 作者索引……(61)

湖南医学院学报
BULLETIN OF HUNAN MEDICAL COLLEGE

前　言

染色体是遗传物质——基因的载体。由于细胞培养和染色体制片及显带技术上的一系列突破，50年代以来人类染色体的研究取得了一系列的重大进展。在我国，由吴旻、项维、刘祖洞等于60年代初最早开展了中国人体的组织培养和外周血培养的染色体研究。70年代初，夏家辉、吴旻等最早引进和建立了染色体显带技术，周宪庭等最早建立了羊水细胞培养技术。1979年李麓芸、夏家辉等开展了定期的遗传咨询门诊服务。全国各地也逐步建立了一支由临床医师和细胞遗传学工作者相结合的遗传咨询队伍，开展了医学细胞遗传学研究和大量临床细胞遗传学工作，在1983年1月召开的“中国遗传学会第二次代表大会”收到的980篇论文中，有203篇（20.7%）属于医学细胞遗传学方面的工作，标志着我国医学细胞遗传学的研究进入了蓬勃发展的新时期。近年来，夏家辉、李麓芸、张思仲、周焕庚、吴旻等开展的高分辨染色体研究，说明我国医学细胞遗传学的研究已取得较大进展，达到了国际先进水平。

为了汇集我国所发现的世界首报染色体异常核型，促进国际间的交流，于1985年9月在湖南医学院医学细胞遗传学国家培训中心举办了第一期研讨会。D.S.Borgaonkar 教授与培训中心主任夏家辉教授、副主任李麓芸副教授组成了一个专门小组，对与会的全国各实验室提交的染色体异常核型标本在分组鉴定、组间互查的基础上，逐个进行了鉴定。确认属于世界首报核型者有185种。这次核实的世界首报核型数目之大，不仅反映了我国人口占世界1/4的特点，而且说明在我国群体中染色体异常的类型是很丰富的。这不但是细胞水平上有关中华民族的一份极有价值的遗传学资料，而且很可能对人类遗传学、肿瘤遗传学及有关的临床医学和基础医学的研究产生深远的影响。因此，与会同志一致要求由夏家辉、李麓芸负责，以这次研讨会的材料为基础，建立“中国人类染色体异常目录”，作为我国人类染色体异常的正式记录。经湖南医学院领导同意，决定由湖南医学院学报编辑部出版第一集。

本书共分两部分，第一部分收载了全国各地84个实验室至1985年8月为止完成染色体检查的人数及所发现的异常核型数。第二部分为经Borgaonkar、夏家辉、李麓芸等鉴定后确认的185种世界首报的异常核型的目录。

需要说明的是，本书第一集收集的资料是参加培训中心第一期研讨会的各单位提供的。未参加这期研讨会的单位的资料没有收入这一集。收入异常核型目录的资料都是经过研讨会反复鉴定的。会后有不少单位又寄来新的资料，由于没有经过鉴定，这次也未收入。这些都将在今后的修订中，通过一定的方式核实后收入。为了

及时总结我国医学细胞遗传学的研究成果，本书拟2—3年增订一次。

由于编者水平有限，加之时间仓促，错漏之处不少，希望同行批评指正。

本书编写过程中得到湖南医学院、湖南医学院学报编辑部和参加研讨会的各单位的大力支持，特此致谢。

编者　85年10月

Preface

Chromosome is the carrier of genes. Since the 1950s the research on human chromosomes has been making a series of progress with the tremendous advances in cell culture, chromosome preparation and banding techniques. In China, the research on human chromosomes using tissue culture and peripheral blood culture was first developed by Wu Min, Xiang Wei, Liu Zudong, et al in the early 1960s. It is the beginning of the research on human and medical cytogenetics in our country. In the early 1970s, the banding techniques of human chromosomes were first introduced by Xia Jiahui, Wu Min, et al. Meanwhile, the amniotic fluid cell culture was first introduced by Zhou Xianting et al. The regular genetic counseling clinic was first set up in Hunan Medical College by Li Luyun and Xia Jiahui in 1979. Since then the ranks of genetic counsellors composed of clinicians and cytogenetists have been all over China. They developed the research on medical cytogenetics and carry out a lot of work in clinical cytogenetics. At the meeting of "The Second Representatives Assembly and Scientific Conference of the Chinese Genetic Association", Jan. 1983, 203 articles of 980 (20.7%) on clinical cytogenetics were received. It illustrated that medical cytogenetics had been used widely in China. In recent years High-Resolution technique of human chromosomes has been researched by Xia Jiahui, Li Luyun, Zhang Sizhong, Zhou Huangen, Wu Min, et al. It showed that medical cytogenetics in China has been in a higher stage and come up to the advanced level of the world.

In order to collect the data of abnormal karyotypes which were first discovered by Chinese cytogenetists in the world and promote the international academic exchanges in our line, "the First National Training Center Conference on Medical Cytogenetics" was held in Changsha in Sept. 1985. Based on the identification of subgroups and groups one by one, a special group which consisted of Professor D.S.Borgaonkar, the chief compiler of "Repository of Chromosomal Variants and Anomalies in Man: An International Registry of Abnormal Karyotypes", Professor Xia Jiahui, the Director of the Training Center and the member of the Scientific Advisory Committee to the Repository-Registry, and Associate Professor Li Luyun, the Vice Director of the Training Center, verified the abnormal karyotypes which were submitted by the participants. They affirmed that there were 185 types of abnormal karyotypes which were first reported in the world. It indicates that there are plenty and various

types of abnormal karyotypes in Chinese population which accounts for one-fourth of the world population. The cytogenetic data are of great value to the Chinese nation and possible to affect the research of human genetics, tumour genetics, and the relevant clinic and basic medicine. Because of this, the participants requested that Xia Jiahui and Li Luyun be in charge of compiling "A Catalogue of Human Chromosomal Anomalies in China", as the formal records of human chromosomal anomalies in China. After the approval of the leader of Hunan Medical College the first collection was published in the name of the Editorial Board of the Bulletin of Hunan Medical College.

The book is composed of two sections. The first section covers the numbers of registered chromosomal analyses and the numbers of abnormal karyotypes which were provided by 84 laboratories in China up to the end of August, 1985. The second section lists 185 types of abnormal karyotypes which were first reported in the world and verified by Professor Borgaonkar, Xia Jiahui and Li Luyun.

The editors deem it necessary to state that only the data received from the participants of "the First National Training Center Conference on Medical Cytogenetics" have been included in the first collection. In other words, this collection does not include the data given by those who did not participate in the conference. The abnormal karyotypes included in the Catalogue were verified during the conference. After the conference, many authors sent us some new data which have not been included in this collection, because they have not been verified. But these data will be included in the revised collection. For the sake of summarizing the achievements in cytogenetic research in China, the book will be supplemented with new materials every two or three years.

The collection has been compiled in haste and the editors' knowledge in the field is limited, so it's bound to have some faults and omissions. We sincerely hope that the readers will oblige us with their valuable comments and criticisms.

We also would like to express our thanks to the leaders of Hunan Medical College, the Editorial Board of the Bulletin of Hunan Medical College and the participants of the conference for their energetic support to this collection.

Compiler 1985.10

GUIDE TO THE USE

使用说明

I. 染色体检查登记：

按提供资料的各实验室名称的汉字笔画顺序排列。

II. 染色体异常目录：

1. 每一核型条目包括以下四部分：

（1）核型描述及病例号，

（2）临床资料，

（3）作者，

（4）引用文献。

凡未注明文献出处者，均仅在医学细胞遗传学国家培训中心第一期研讨会上报告，并经过鉴定。

2. 核型条目按序编排，编号的头两位数字指染色体（01—22，OX，OY）；第三位指染色体臂（p,q）；第 4、5 位数字分别指区和带。如能将断裂点定位于一个染色带内，则按 ISCN (1978)的建议，将这个带分成10个单位，并根据断裂点到该带近侧缘的相对距离，将其记入第6、7位。例如，06p2105指距06p21带的近侧缘5/10处的断裂。若断裂点可定位到亚带，则按ISCN(1978,1981)关于带的再分法的规定编号。例如 01p36.11，小数点后的两位数字表示01p36带的亚带，即1号染色体短臂3区6带第 1 亚带第 1 次亚带。当异常核型涉及两个或更多断裂点（或异常）时，则登记两处，并在第二个断裂点（或异常）的条目下加以说明。例如 t(6；21)(p25；q11)在06p250有其完整条目而在 21q110 条目下注明：同样条目见06p250。

3. 核型条目采用 ISCN(1978、1981)制定的术语和缩写符号，按繁式体系描述。

4. 目录的每一页上附有取自 ISCN(1981) 模式图的染色体图，以帮助读者理解在这一页上描述的染色体区和带。

5. 为节约编幅，本目录全部用英语描述，未加中文说明。

III. 书末附录为作者索引，包括作者姓名和工作单位，按作者姓名的汉语拼音字母顺序排列。

Guide to the Use

I. Registry of Chromosomal Examination:

In the table the data are arranged in the order of the number of strokes of the Chinese Character of the laboratory's names which afforded the information.

II. Catalogue of Chromosomal Anomalies:

1. In the Catalogue every entry consists of four parts:

(1) karyotype and the subject number in the laboratory

(2) clinical information

(3) authors

(4) reference

All of the karyotypes which were not indicated the references were reported and identified at "The First National Training Center Conference on Medical Cytogenetics".

2. In the Catalog the entry number are arranged numerrically, the first two digits refer to the chromosome numbers (01 to 22, OX and OY); the third digit to the chromosome arms (p and q); and the fourth and fifth digits to the region and band. When a break point can be specified within a band, the band will be divided into 10 units according to the suggestion of ISCN(1978), and the information will be entered in the sixth and seventh digits on the basis of the relative distance of the break point from the proximal margin of the band concerned. For examp'e, 06p2105 implies a break halfway from the proximal edge of band 06p21. If a break point can be located in a sub-band, it will be arranged according to the method for subdividing bands of ISCN (1978; 1981). For example, 01p36.11, the two digits after the punctuation indicate the sub-bands of 01p36. Whenever a karyotype has information on two or more break points (or Abnormalities), an appropriate comment is at the second point of entry. For example, the translocation t(6; 21)(p25; q11) will have its complete entry at 06p250, and at 21q110 its secondary entry will have the following notation: same entry as in 06p250.

3. the entries are discribed using the Detailed System with the terms and abbreviations recommended by ISCN (1978; 1981).

4. On each page there are one or more pictures of the relevant chromosomes from the ISCN (1981) ideogram. This will help in the understanding of the chromosome regions and bands that are being discussed on that page.

III. Appendix at the end of the book is an Author Index. It includs the names and addresses of the authors and is arranged in the order of the number of Chinese Phonetic Alphabet in the author's surnames.

I. REGISTRY OF CHROMOSOMAL EXAMINATION

染色体检查登记

(up to the end of August, 1985)

(截至1985年8月)

染色体检查登记

（截至1985年8月）

Registry of Chromosomal Examination

(up to the end of August, 1985)

Name of The Lab 单　　位	Total 染色体检查 总人数	Anomalies 染色体异常 人　数
广东省计划生育研究所 遗传室 Genet Lab Guangdong Institute of Family Planning Guangzhou, Guangdong, 777331	150	11
广西壮族自治区人民医院 遗传研究室 Med Genet Lab Guangxi People's Hosp Nanning, Guangxi, China	410	24
广西医学院 医学遗传研究室 Med Genet Lab Guangxi Med Coll Nanning, Guangxi, China. 31477	467	30
广西壮族自治区妇幼保健院遗传室 Med Genet Lab Women and Children's Hosp Nanning, Guangxi, China　20693	453	36
上海市儿童医院医学遗传研究室 Med Genet Lab Shanghai Children's Hosp Shanghai, China　539696	341	53
上海市第二医科大学医学遗传学教研室、细胞遗传实验室 Cytogenet Lab Dept of Med Genet Shanghai 2nd Med Univ Shanghai, China　260760	378	139
上海市第六人民医院 医学遗传研究室 Med Genet Lab The 6th People's Hosp Shanghai, China	1606	180

Name of The Lab 单　　位	Total 染色体检查 总人数	Anomalies 染色体异常 人　数
上海市第六人民医院妇产科病理研究室 Obstet Patho Lab The 6th People's Hosp Shanghai, China	303	10
上海市静安区中心医院、细胞遗传室 Cytogenet Lab Jingan District Central Hosp Shanghai, China 530109	1519	141
山东医科大学、生物教研组、医学遗传研究室 Med Genet Lab Dept of Biology Shandong Med Univ Jinan, Shandong, China 26052	463	71
山东医科大学 附属医院、小儿科细胞遗传实验室 Cytogenet Lab, Dept of Pediatrics The Affiliated Hosp, Shandong Med Univ Jinan, Shandong	936	75
山东省淄博市、妇幼保健院遗传实验室 Med Cytogenet Lab Maternal & Children health Hosp Zibo, Shandong, China 33991	630	15
山西省大同市第三人民医院遗传室 Med Genet Lab The 3rd People's Hosp Datong, Shanxi, China	112	11
山西医学院第一附属医院妇产科、计划生育研究室 Family Planning Lab Dept of Obstet & Gynec The 1st Affiliated Hosp Shanxi Med Coll Taiyuan, Shanxi, China 22114	640	37
山西医学院第二附属医院妇产科遗传室 Med Genet Lab Dept of Gynec & Obstet The 2nd Affiliated Hosp Shanxi Med Coll Taiyuan, Shanxi.	560	28

Name of The Lab 单　　位	Total 染色体检查 总人数	Anomalies 染色体异常 人　数
山西省妇幼保健院医学遗传室 Med Genet Lab Shanxi Obstet & Gynec Hosp Taiyun, Shanxi, China 28116	389	36
大连医学院附属医院妇产科遗传研究室 Genet Lab Dept of Obstet & Gynec The 1st Affiliated Hosp Dalian Med Coll Dalian, Liaoning, China	1128	50
天津市工人医院 天津市河东医院、计划生育技术指导所、遗传实验室 Tianjin Worker's Hosp Med Genet Lab Family Planning Guide Center Tianjin Hedong Hosp Tianjin, China	364	28
天津市中心妇产科医院遗传室 Med Genet Lab Tianjin Central Hosp of Obstet & Gynec Tianjin,	987	54
中山医科大学、第一附属医院妇产科、细胞遗传室 Cytogenet Lab Dept of Gynemetrics The 1st Affiliated Hosp Zhong Shan Med Univ Guangzhou, Guangdong	958	74
中山医科大学第二附属医院妇产科、染色体实验室 Cytogenet Lab Dept of Gynemetrics The 2nd Affiliated Hosp Zhong shan Med Univ Guangzhou, Guangdong	737	43

Name of The Lab 单　　位	Total 染色体检查 总人数	Anomalies 染色体异常 人　数
中国人民解放军总医院妇产科实验室 Med Genet Lab Dept of Obstet & Gynec P L A General Hosp Beijing	295	20
中国人民解放军第四军医大学第一附属医院妇产科实验室 Genet Lab Dept of Obstet & Gynec The 1st Affiliated Hosp The 4th Military Med Univ Xian, Shanxi	1155	62
中国人民解放军，第408医院医学遗传实验室 Med Genet Lab Hosp of the PLA No 408 Qinhuangdao Hebei	211	12
中国医科大学附属一医院儿科遗传实验室 Pediatrics Lab The 1st Affiliated Hosp China Med Univ Shenyang, Liaoning, China	1388	122
中国医科大学附属二医院小儿科细胞遗传室 Cytogenet Lab Dept of Pediatrics The 2nd Affiliated Hosp China Med Univ Shenyang, Liaoning, China	1323	284
中国医科大学附属三医院、医学细胞遗传实验室 Med Cytogenet Lab Dept of Obstet & Gynec The 3rd Affiliated Hosp China Med Univ Shenyang, Liaoning, China	2258	102
云南省第一人民医院妇产科实验室 Cytogenet Lab Dept of Obstet & Gynec The 1st People's Hosp of Yunnan Prov Kunming, Yunnan, China	457	43

Name of The Lab 单　　位	Total 染色体检查 总人数	Anomalies 染色体异常 人　数
内蒙古自治区医院、妇产科医学细胞遗传室 Med Cytogenet Lab Dept of Obstet & Gynec Inner Mongolian Autonomous Region Hosp Huhehot, Inner Mongolia, China 43631	351	8
内蒙古医学院、附属医院、妇产科细胞遗传室 Cytogenet Lab Dept of Obstet & Gynec The Affiliated Hosp Inner Mongolia Med Coll Huhehot, Inner Mongolia, China	535	14
兰州医学院第一附属医院、妇产科医学遗传实验室 Med Genet Lab Dept of Obstet & Gynec The 1st Affiliated Hosp Lanzhou Med Coll Lanzhou, Gansu, China	450	19
兰州医学院第二附属医院妇产科实验室 Obstet & Gynec Lab The 2nd Affiliated Hosp Lanzhou Med Coll Lanzhou, Gansu, China.	856	23
甘肃省妇幼保健院 The Maternity & Children Health Hosp of Gansu Lanzhou, Gansu, China.	556	15
四川省成都市妇产科医院 The Obstet & Gynec Hosp Chendu, Sichuan, China	515	32
白求恩医科大学第二临床学院、妇产科遗传实验室 Genet Lab Dept of Obstet & Gynec The 2nd Affiliated Hosp N. Bethune Univ of Med Sciences Changchun, Jilin, 64617	296	12