Karyotype

Repository of Human Chromosomal Anomalies In China

●主　编:夏家辉
First Chief Editor: XiaJiahui

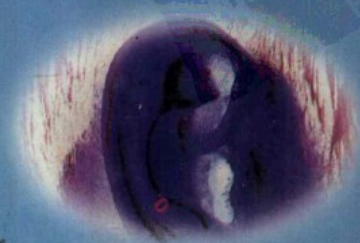
Clinic Data

中国人类染色体异常核型数据库

Repository of Human Chromosomal Anomalies In China

Reference

Source



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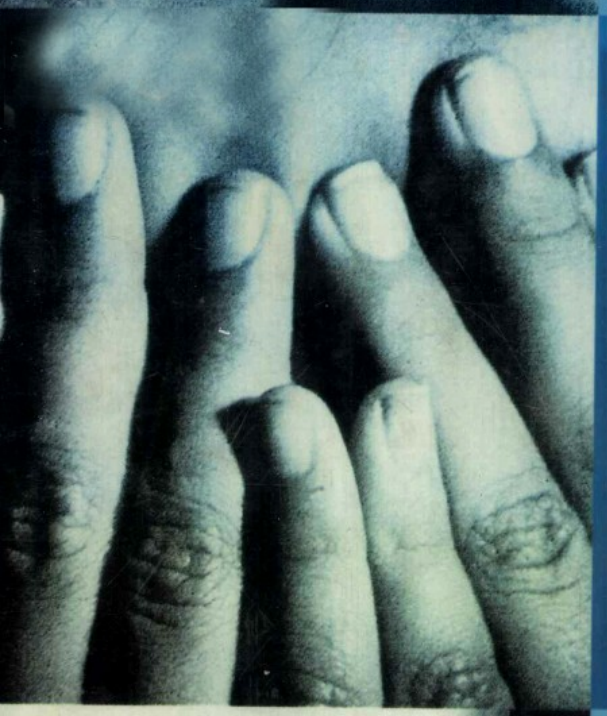

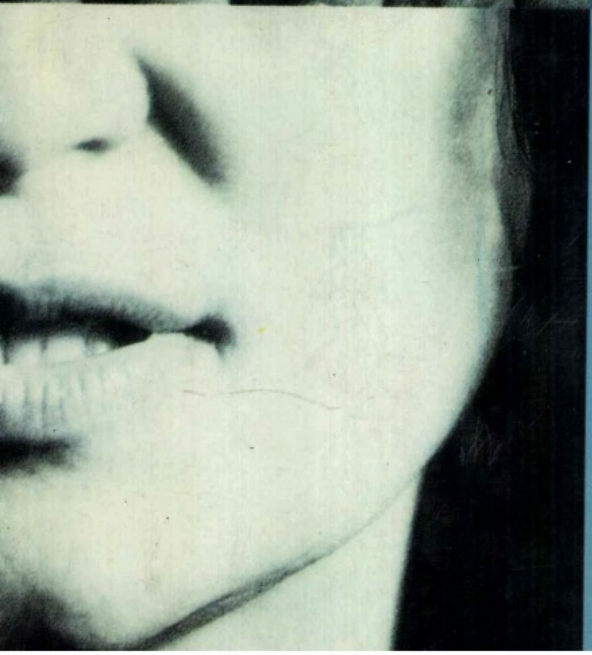

中国人类染色体异常核型数据库

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中国人类染色体异常核型数据库

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湖南科学技术出版社

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序

本数据库曾以《湖南医科大学学报》专辑于1985年和1990年出了二版，有关内容均已转入美国 D. S. Borgaonkar 主编的《Repository of Human Chromosomal Variants and Anomalies — An International Registry of Abnormal Karyotypes》一书中。现将我们自1985年至1996年6月底所收集、鉴定的来自全国257个实验室658位细胞遗传学工作者所发现的世界首报核型1210种编辑成册，并由湖南科学技术出版社正式出版，以促进我国医学遗传学工作者与国际间的交流。

医学遗传学国家重点实验室
学术委员会主任、博士导师 **夏家辉**
第一主编
1996年7月

The Preface of The Third Edition

The first and second edition of this catalogue《A CATALOGUE OF HUMAN CHROMOSOMAL ANOMALIES IN CHINA》was published as special editions of Bulletin of Hunan Medical University. The contents were reprinted in《Repository of Human Chromosomal Variants and Anomalies — An International Registry of Abnormal Karyotypes》. Now, we compiled all the data of first reported abnormal karyotypes in the world we collected and verified which were provided by 658 cytogenetists of 257 laboratories in China and published it by Hunan Science and Technology Press. We hope this work can enhance the communication between Chinese and foreign medical geneticist.

Chairman of Academic Committee of
National Lab of Medical Genetics
Advisor of Doctor, First Chief Editor
Xia Jiahui
July, 1996

湖南医学院学报 1990 年专辑序

1985 年出版的《中国人类染色体异常目录》，不但得到了国内同行的高度评价与支持，而且于 1987 年在美国 D. S. Borgaonkar 主编的《Repository of Human Chromosomal Variants and Anomalies — An International Registry of Abnormal Karyotypes》一书的第十二版中全部作了转载，说明该目录已达到了国内外交流的目的。近年来，我国的研究工作又有新的进展，截至 1990 年 12 月底，我们又收到国际目录中未曾记载的核型 547 种，累计核型 732 种，这些核型来自我国 189 个实验室的 470 位细胞遗传学工作者。现特编辑成第二版，并全部储存在计算机中，以便查询和进一步扩编。

夏家辉　李麓芸
1990 年 12 月于湖南医科大学
医学遗传学国家重点实验室

The Preface of The Second Edition

The 1st edition of "A Catalogue of Human Chromosomal Anomalies in China" was publiched in 1985. It was not only supported and evaluated highly by the colleagues in China, but also was reprinted in《Repository of Human chromosomal Variants and Anomalies — An International Registry of Abnomal Karyotype》, the 12th edition in 1987, which was edited by D. S. Borgaonkar in USA, showing that the 1st edition had achieved the goal of communicating with colleagues both in China and abroad.

Recent years, new approaches of the work had made in our country till the end of Dec. 1990. We had received 547 karyotypes which were not recorded by " Repository of Human Chromosomal Variants and Anomalies". Total kinds of karyotypes were 732 that were from 470 cytogenetists of 189 labs. We have compiled the 2nd edition of the catalogue and have established "The Data Bank of Human Chromosomal Anomalies in China" for storing and exchanging. It will faciliate the communicating wider in the future.

Xia Jiahui
Li Luyun
The State Key Lab of Medical Genetics of China
Hunan Medical University
Changsha, Hunan, China
Dec. 1990

湖南医学院学报 1985 年专辑前言

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，周先庭等最早建立了羊水细胞培养技术。1979 年李龑芸、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的研究和大量临床细胞遗传学的工作，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、李龑芸、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了较大进展，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、李龑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、李龑芸负责，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、李龑芸等鉴定后确认的 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

1985 年 10 月

The Preface of The First Edition

Chromosome is the carrier of genes. Since the 1950s the research on human chromosome 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 cultrue was fisrt 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 frist 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 cytogenetist 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 Huangeng, 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 cytogenetist in the world and promote the international acadmic exchanges in our line, "the First National Training Center Conference on Medical Cytogenetics" was held in Changsha in Sept. 1985. Based on the identificationa of subgroups and groups one by one, a special group which consisted of Professor. S. Borgaonkar, the chief compiler of "Repository of Chromosomal Variants and Anomalies in Man: An International Registry of Abnormal Karyotype", 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 abmornal 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 secitions. 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 include in the frist 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 cytogenetics 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 University, the Editorial Board of the Bulletin of Hunan Medical College and the participants of the conference for their energetic support to this collection.

Complier 1985. 10

目　录

47，XY，fra（X）

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：47，XY，fra（X）.

Clinic Data A 13-year-old boy，fat. He had mental retardation.

Source Xia Jiahui，Li Luyun，Dai Heping，Long Zhigao

National Lab of Medical Genetics，Hunan Medical University，Changsha，Hunan 410078

夏家辉　李麓芸　戴和平　龙志高

湖南医科大学医学遗传学国家重点实验室　邮政编码：410078

45，XY，－21/46，XY/47，XY，＋14

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：45，XY，－21/46，XY/47，XY，＋14.

Clinic Data A 6-year-old with mental retardation. G1 P1，weight of birth：2.95kg. He couldn't raise his head at 1-year-old，and could not walk and speak at 2 1/2-year-old. Physical examination：height 98.5cm，weight 15.5kg，head circumference 52.8cm. He had developmental retardation，dry skin with brown macula，frontal slanting，prominent occiput，hypertelorism，broad base of nose，wide mouth，yellow teeth which in bad order，micrognathia and retrognathia，large and lowset ears，short neck and pigeon breast，IQ 54.

Source Song Xiaomin

Genetic Lab of Hunan Children's Hospital，Changsha，Hunan 420031

宋晓敏

湖南省长沙市，湖南省儿童医院遗传室　邮政编码：420031

47，XY，＋21/94，XXYY，＋21，＋21

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：47，XY，＋21/94，XXYY，＋21，＋21.

Clinic Data A 8-month-old boy with mental retardation and underdeveloped，Height 63cm，Weight 6kg，short neck，flat forehead，microphthalmia，hypertelorism，external canthus was pulled downward，flat nasal bridge，small mouth and thick lips，tongue extends out，simian crease on left hand.

Source Wang Zhaocai，Huang Feifei

Eugenics Laboratory，Henan Provincial People's Hospital，Zhengzhou，Henan 450003

王兆才　黄飞飞

河南省郑州市，河南省人民医院优生研究室　邮政编码：450003

46，XX/46，XY/47，XXX/47，XXY.

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：46，XX/46，XY/47，XXX/47，XXY.

Clinic Data A 36-year-old female，carrier. She had 2 spontaneous abortions which occurred in the 2nd to 4th months of pregnancy.

Source Wang Yingtai，Li Congmin，Peng Suwen

Genetics Lab，Henan Institute of Family Planning，Zhengzhou，Henan 450003

王应太　李聪敏　彭素文

河南省计划生育科研所遗传室　邮政编码：450003

+9

45, X/46, XX/47, XX, +9

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 45, X/46, XX/47, XX, +9.

Clinic Data A 26-year-old female, carrier. She had given birth to 2 babies with malformations, and all died at the 4th day after birth.

Source Wang Yingtai, Wang Zhaocai, Yang Yanli, Hao Gefang. Eugenics Lab, The People's Hospital, Henan, Scientific Research Institute of Family Planning of Henan 450003

王应太　王兆才　杨艳丽　郝戈芳

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+mar

45, X/46, XX/46, X, +mar

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 45, X/46, XX/46, X, +mar.

Clinic Data The patient is an 18-year-old female. She has short stature, mental retardation, underdevelopment of breasts, primary amenorrhea and malformations primarily including cubitus and webbing of the neck. Her parents are first cousins. Her parents and sibs are all normal.

Source Gu mingmin, Shen Ruocui, Zhang Lijun, Su Yubin

Department of Medical Genetics, the 2nd Shanghai Medical University, Shanghai 200025

顾鸣敏　沈若崔　张丽君　苏宇滨

上海第二医科大学遗传学教研室邮政编码：200025

01cen

46, XY, t (1; 9) (1p9p; 1q9q)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XY, t (1; 9) (1pter→cen→9pter; 9qter→cen→1qter).

Clinic Data A 32-year-old male, carrier. His wife had 2 spontaneous abortions which occurred in 1st trimester of pregnancy.

Source Zheng Jun, Zeng Fen, Yang Liu, Wei Caiyue

Medical Genetics Lab, The Women and Children's Health Hospital of Shaanxi Prov, Xian, Shaanxi 710003

郑　军　曾　芬　杨　柳　韦彩月

陕西省西安市，陕西省妇幼保健院医学遗传实验室　邮政编码：710003

46, XX, t (1; 10) (1q10p; 10q1p)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XX, t (1; 10) (1qter→1cen→10pter; 10qter→10cen→1pter).

Clinic Data The 30-year-old female, She had primary infertility and masculinization.

Source Du Jing, Li Xiulin, Li Li

Medical Cytogenetics Lab, Department of Pediatrics, the 1st Affiliated Hospital, Chinese Medical University, Shenyang, Liaoning 110000
杜 晶　李修林　李 莉
辽宁省沈阳市，中国医科大学附一院小儿科遗传室 邮政编码：110001

01p11

46, XX t (1; 12; 22; 15; 11; 8) (p11; q11; p11; q15; q21; p23)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XX, t (1; 12; 22; 15; 11; 8) (1qter→1p11::8p23→8pter; 12pter→12q11::1p11→1pter; 22qter→22p11::12q11→12qter; 15pter→15q15::22p11→22pter; 11pter→11q21::15q15→15qter; 8qter→8p23::11q21→11qter).

Clinic Data A 28-year-old female, carrier. She had 3 spontaneous abortions which all occurred in the 2nd month of pregnancy.

Source Wu Subin
Cytogenetics Lab, Department of Gynecology and Obstetrics, the 1st Affiliated Hospital of Zhongshan Medical University, Guangzhou, Guangdong 510080
吴素彬
广东省广州市，中山医科大学附一院妇产科细胞遗传室　邮政编码：510080

46, XX, t (1; 19) (p11; p13)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XX, t (1; 19) (1qter→1p11::19p13→19pter; 19qter→19p13::1p11→1pter).

Clinic Data A 28-year-old female, carrier. She had a baby with encephalocele and died after birth.

Source Ding Yan, Wang Xiqin, Li Jinhua, Guo Zhanying
Family Planning Lab, the 1st Affiliated Hospital of Shanxi Medical College, Taiyuan, Shanxi 030000
丁 琰　王希琴　李晋华　郭湛英
山西省太原市，山西医学院附一院计划生育研究室　邮政编码：030000

01p12

46, XX, t (1; 6) (p12; q15)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XX, t (1; 6) (1qter→1p12::6q15→6qter; 6pter→6q15::1p12→1pter).

Clinic Data A 28-year-old female, carrier. She had 1 spontaneous abortion.

Source Ruan Yiping, Zhu Kunyi, Li Dinghua
Genetic Lab, Women and Children's Health Hospital, Jiangmen, Guangdong 529020
阮一平　朱坤仪　李定华
广东省江门市妇幼保健院遗传室　邮政编码：529020

46, XY, t (1; 9) (p12; q13)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XY, t (1; 9) (1qter→1p12::9q13→9qter; 9pter→9q13::1p12→1pter).

Clinic Data A 33-year-old male. His wife had 2 spontaneous abortions.

Source Xu Yimin

Genetics Lab, Fuxing Hospital, Beijing 100038

许怡民

北京市复兴医院遗传室　　邮政编码：100038

46, XX t (1; 19) (p12; p13)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XX, t (1; 19) (1qter→1p12::19p13→19pter; 19qter→19p13::1p12→1pter).

Clinic Data A 2-year-old girl with physical and mental retardations.

Source Wang Xinghan, Zhang Qiliang

Medical Genetic Lab, People's hospital of Weifang, Weifang, Shandong 261041

王兴汉　　张奇亮

山东省潍坊市，人民医院遗传室　　邮政编码：261041

01p13

46, XY, t (1; 7) (p13.1; p11.2)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XY, t (1; 7) (1qter→1p13.1::7p11.2→7pter; 7qter→7p11.2::1p13.1→1pter).

Clinic Data A 4-year-old boy with mental and growth retardation.

Source Tang Yingjie

Test department of Longquanshan Hospital, Guangxi 530000

汤英杰

广西龙泉山医院检验科　　邮政编码：530000

46, XX, t (1; 7) (p13; q36)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XX, t (1; 7) (1qter→1p13::7q36→7qter; 7pter→7q36::1p13→1pter).

Clinic Data A 30-year-old female, carrier. She had 2 spontaneous abortions which occurred in the 2nd month of pregnancy.

Source Xu Yimin

Genetics Lab, Fuxing Hospital, Beijing 100038

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北京市复兴医院遗传室　　邮政编码：100038

46, XX, t (1; 16) (p13; q13)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XX, t (1; 16) (1qter→1p13::16q13→16qter; 16pter→16q13::1p13→1pter).

Clinic Data Female, carrier.

Source Liu Linwei

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46，XY t（1；20）（p13；q13）pat

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：46，XY，t（1；20）（1qter→1p13::20q13→20qter；20pter→20q13::1p13→1pter）pat.

Clinic Data A 30-year-old male，carrier. His wife had a full term abnormal baby and 2 spontaneous abortions in the 1st trimester of pregnancy. Then his wife delivered a child whose phenotype was normal，but Karyotype was the same as his father.

Source Shen Ruduan，Gao Hua，Zhang Lan
Cytogenetic Lab，Department of Obstetrics and Gynecology，the 1st Affiliated Hospital of Kunming Medical College，Kunming，Yunnan 650032
沈汝端　高　桦　张　兰
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01p22

46，XX，inv（1）（p22p36），inv（9）（p13q13）

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：46，XX，inv（1）（pter→p22::p36→p22::p36→qter），inv（9）（pter→p13::q13→p13::q13→qter）.

Clinic Data A 21-year-old female. She had mental retardation，and orbital hypertelorism. webbing of the neck，simian arease，short small fingers. X-ray：Her heart is on right. Her father karyotype's is：46，XY，inv（1）（p22；p36），her mother's karyotype is：46，XX，inv（9）（p13；q13）.

Source Li Wendian，Gong Wuxing，Zhang Xuhua
Women and Children's Health Hospital of Zhuhai，Guandong 519000
李文典　龚五星　张旭华
广东省，珠海市妇幼保健院邮政编码：519000

46，XY，t（1；2）（p22；q37）

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：46，XY，t（1；2）（1qter→1p22::2q37→2qter；2pter→2q37::1p22→1pter）.

Clinic Data A 32-year-old male with primary sterility.

Source Zhang Baozhen
Genetics Lab，Fuzhou General Hospital of Nanjing Military Region，Fuzhou，Fujian 350001
张宝珍
福州南京军区福州总院遗传室　　邮政编码：350001

46，XX，t（1；3）（p22；p11）

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：46，XX，t（1；3）（1qter→1p22::3p11→3pter；3qter→3p11::1p22→1pter）.

Clinic Data A 24-year-old female. She had a secondary amenorrhea.

Source Wang Qin，Chen Zhangyu
Medical Cytogenetic Lab，Number 202 Hospital，Shenyang，Liaoning 110000

王 勤　陈章玉
辽宁省沈阳市，沈阳第 202 医院遗传室　邮政编码：110000

46，XY，t（1；3）（p22；p25）

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：46，XY，t（1；3）（1qter→1p22::3p25→3pter；3qter→3p25::1p22→1pter）.

Clinic Data A 28-year-old male. His wife had 1 abortion history occurred in the 3rd month of pregnancy.

Source Xu Yimin
Genetics Lab，Fuxing Hospital，Beijing 100038
许怡民
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46，XY，t（1；3）（p22；q21）

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：46，XY，t（1；3）（1qter→1p22::3q21→3qter；3pter→3q21::1p22→1pter）.

Clinic Data A 31-year-old male，carrier. His wife had 2 spontaneous abortions which occurred in the 1st trimester of pregnancy.

Source Wang Aiqin，He Chunna，Zhang Haiduan
Genetics Lab，The Air Force Hospital of Nanjing，Nanjing，Jiangsu 210002
王爱勤　何春娜　张海端
江苏省南京市，南京空军医院遗传室　邮政编码：210002

46，XX，t（1；4；3）（p22；p16；q21）

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：46，XX，t（1；4；3）（1qter→1p22::3q21→3qter；4qter→4p16::1p22→1pter；3pter→3q21::4p16→4pter）.

Clinic Data A 9 year-old girl was born at full term and was the 1st child，with mental retardation，height 115cm.

Source Ma Renyi
Leshan People's Hospital，Leshan，Sichuan 614000
马仁义
四川乐山市人民医院　邮政编码：614000

46，XX，t（1；4）（p22；q21）

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is：46，XX，t（1；4）（1qter→1p22::4q21→4qter；4pter→4q21::1p22→1pter）.

Clinic Data The female aged 39，with normal phenotype，had three pregnancies，two of which ended in spontaneous abortions at about 3 months of gestation. The single child alive is a healthy girl. His husband's karyotype was normal.

Source Li Yongquan
Lab of Medical Genetics，Guangdong Medical College 524023
李永全
广东医学院医学遗传学研究室　邮政编码：524023

46, XX, t (1; 4) (p22; q27) mat

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XX, t (1; 4) (1qter→1p22::4q27→4qter; 4pter→4q27::1p22→1pter) mat.

Clinic Data A new-born with congenital wryneck.

Source Xie Zhimin

Department of Obstetrics and Gynecology, Maternal Children Health Hospital, Western City District. Beijing 100044

谢志敏

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46, XX, t (1; 4) (p22; q31) mat

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XX, t (1; 4) (1qter→1p22::4q31→4qter; 4pter→4q31::1p22→1pter) mat.

Clinic Data A 3-month-old girl. She had an oncoides in her left labia. Her mother, aunt and her uncle had the history of spontaneous abortions and the same translocation chromosomes as the proband.

Source Zhu Junzhen, Hao Yubin, Li Hongyun, Yu Xiaoping

Medical Genetic Lab, Hebei Provincial Hospital, Shijiazhuang, Heibei 050071

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46, XX, t (1; 7) (p2205; q3600)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XX, t (1; 7) (1qter→1p2205::7q3600→7qter; 7pter→7q3600::1p2205→1pter).

Clinic Data A 26-year-old female, carrier. She had 4 spontaneous abortions which occurred in the 1st trimester of pregnancy.

Source Li Luyun, Xia Jiahui, Dai Heping, Xu Faming, He Xiaoxuan, Xu Jia

Medical Genetics Lab, Hunan Medical University, Changsha, Hunan 410078

Reference: Chinese Medical Journal 1986; 99 (7): 527～534.

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中华医学杂志 英文版　1986; 99 (7): 527～534

46, XY, t (1; 8) (p22; q1309)

Karyotype The G-banded chromosomes of peripheral blood lymphocytes analysis show that the karyotype is: 46, XY, t (1; 8) (1qter→1p22::8q1309→8qter, 8pter→8q1309::1p22→1pter).

Clinic Data A 32-year-old male, carrier. His wife had 2 dead infants.

Source Xia Jiahui, Li Luyun, Dai Heping, Long Zhigao

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46, XX, −1, −11, +der (1) t (1; 11) (1qter→1q21::1p22→cen::11p13→11pter), +der (11) t (1; 11) (11qter→cen::1q11→1q21::1p22→1pter)