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复旦大学遗传学研究所

年度报告

Annual Report July, 1982—December, 1983

INSTITUTE OF GENETICS FUDAN UNIVERSITY

SHANGHAI

PEOPLE'S REPUBLIC OF CHINA

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宝山大学遗传学研究所

年度报告

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前　言

复旦大学遗传学研究所1982～1983年度的研究报告现在和读者见面了。这是我所自1979年以来正式汇编的第四期年报。在这一年中共接受国家科委重点课题七项，中科院科学基金会三项，教育部下达课题三项，上海市科委课题四项和其他兄弟单位协作课题四项。从本期收录的自1982年6月至1983年12月的53篇摘要中可以看到这些工作的某些进展情况。

除了这些研究工作之外，在这一年中，我所还参加了一些其他活动。例如1983年1月在福州市举行的第二届全国遗传学会年会。由我所提交给大会的论文共42篇。有三名出国进修人员在大会上作了工作汇报。同年5月受中国遗传学会委托和上海市遗传学会一起联合召开国际三致(致癌、致变、致畸)学术会议，应邀来沪与会的有来自美、日、英、法等国的外国学者、教授共35名，国内专家120名。我所参加了该国际会议的组织工作，宣读论文5篇。6月份我所委派盛祖嘉教授出席在日本东京召开的国际工业微生物遗传学会议。7月份刘祖洞教授受教育部委托赴意大利进行教育考察。12月份以所长谈家桢教授为团长的中国遗传学代表团出席了在印度召开的国际遗传学会第15届年会。我所提交给大会的学术论文有4篇，作为代表团成员赴会的有教授、副教授三人。

在国际协作方面除经常接待各国访问学者作学术报告外，美国康奈尔大学吴瑞教授、日本东京都立大学北川修和冈田两位教授曾先后应邀来我所作系统讲学，并就水稻基因文库，水稻雄性不育三系的分

子机理以及果蝇分类等课题进行国际协作。

本年度报告在编辑出版过程中得到了上海《世界科学》杂志的支持和协助。谨此一并致谢。

复旦大学遗传学研究所

1983年12月

PREFACE

The annual report (from June 1982 to December 1983) has now been available to our readers. As you may see, this report is the fourth of its kind since the first was published in 1979. During this period (from June 1982 to December 1983) studies were carried out on 7 important research projects supported by grant from the Chinese State Commision for Science and Technology , 3 from Foundation of Chinese Academy of Sciences, 2 from the Ministry of Education, 4 from Shanghai Commissian for Science and Technology and 4 in collaboration with other institutes. Our readers will be invited to read these 53 abstracts which reflect the progress we have made in these fields.

Apart from these research programmes, our institute was also actively engaged in other scientific activities. For instances, a total of 22 papers were submitted to the 2nd annual meeting of The Chinese Genetics Society held in January 1983. Also in this annual meeting, 3 of our staff members who had just returned from abroad reported on their research work in countries they had visited. In May 1983 an International Workshop on Environmental Mutagenesis, Carcinogenesis and Teratogenesis was held in Shanghai, attended by 35 prominent scientists from abroad and 120 Chinese specialists. The workshop was co-sponsored by The Chinese Genetics Society and the Internation Environmentol Mutagen Society. Our institute was responsible for the organization work. In this meeting 5 papers were presented which dealt with the research work carried out in our institute. In June 1983, Professor Sheng Zujia was invited to attend the 5th International Industrial Microbial Genetics Conference in Tokyo, Japan. In July, Professor Liu Zudong entrusted by the Ministry of Education went to Italy for an inspection tour. In December 1983 Professor C. C Tan, director of our institute, together with 3 senior researchers, attended the XV International Congress of Genetic held in New Delhi. Professor Tan acted as the vice-chairman of the Congress and the head

of Chinese Delegation. In this Congress 4 papers were submitted by our institute.

This year also witnessed fruitful scientific exchanges between our staff members and visiting scholars from many countries. Professor Wu Rui of Cornell Uninversity, Professor Kitakawa and Professor Okata of Tokyo Metropoliton Uniersity were invited to give a series of lectures in our institute and international collaboration on such research projects as gene library of rice, molecular mechanism of male sterile system in rice and systematic studies on Drosophila in China initiated.

Institute of Genetics, Fudan University

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一、人类和医学遗传学研究室

1982年7月以来，我室接受中国科学院科学基金资助项目研究工作，课题名称为：人体基因组在正常和异常情况下的结构、调控与表达。1983年下半年又接受国家科委细胞生物学基础研究的项目，课题名称为：人类基因定位和基因调控的研究。在六五期间，我们的目标是建立无性杂交系分布板，人的第12号染色体的DNA文库，用DNA和小细胞转移技术研究基因转移，基因表达和基因调控。1983年5月在上海召开的国际致突变、致癌和致畸讲习会上，介绍了我室近年来在致突变研究方面的工作。1983年12月项维付教授出席在印度新德里召开的第15届国际遗传学会议，并宣读了论文。1983年我室为本科大学生开设了体细胞遗传学课程，接受兄弟院校进修教师共14名，今年又新招收3名硕士研究生，现我室共有硕士研究生8名。本年度内尚有多批教师参加这种全国培训班和各种学会的年会活动。並新派出3名教师去美国和西德进修学习。

Section of Human and Medical Genetics

Since July 1982, we have been working on the research project entitled "The structure and function of the normal and diseased human genome" supported by grant from Science Foundation. In Late 1983, we got another grant from National Association for Science and Technology to carry on research work on "Human gene mapping and gene regulations". Our research target for the coming 3 years is to establish a human and rodent hybrid cell clone panel, the DNA library from single human chromosome 12. DNA and microcell mediated gene transfer, gene expression and gene regulation. In May 1983, The International Workshop on Environmental Mutagenesis, Carcinogenesis and Teratogenesis was held in Shanghai. In this meeting we reported our research progress on environmental mutagenesis. In December 1983, Professor Xiang Wei attended the XV International Congress of Genetics held in New Delhi, India. We submitted to the Congress an abstract concerning about the mutagenicity of the gossypol. In 1983, we offered a course on the somatic cell genetics for the senior students. we had trained 14 advanced teachers this year from various colleges, universities and hospitals all over the country. We have totally 8 students who are working for master degree (including 3 of them were receiving this year). We sent another 3 staff members to US and West Germany for advanced training.

(一)研究工作(Research Works)

1—1 脆性X染色体综合症(附一例报告)

薛京伦　刘　欣*　俞民澍
谈延德**　潘　犁　邱信芳
朱畅宁***　冯玲英***

本文报告脆性X染色体综合症一例。脆性X染色体综合症系一种非特异性X连锁智力低下。本例男孩临床表现为棕黄色头发和耳大等异常体征，其总发育商（DQ）为80，比正常同龄儿童的DQ低8个月。患儿外周血淋巴细胞培养，经同步化处理后用Giemsa染色，显示脆性X染色体。本文还就脆性X染色体检测的临床意义，细胞遗传学特征与产生机制，以及检测方法等有关问题作了简要讨论。

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Fragile X Syndrome (A Case Report)

Xue Jinglun　Liu Xin　Yu Minshu　Tan Yande　Pan Li
Qiu Xinfang　Zhu Changning　Fen Lingying

A mentally retarded boy, aged 3, with brown hair and macrotia, of fragile X syndrome is reported. Fragile X syndrome is one of the non-specific X-linked mental retardations. The total development quotient (DQ) of this patient was 80, 8 months less than that of normal boy. A fragile X was demonstrated by chromosome analysis with Giemsa stain after synchronization of the patient's blood lymphocyte cultured. In addition, the clinic value, cytogenetic characteristics and method for demonstration of fragile X were briefly discussed.

1—2 地方性克汀病患者与正常人尿液中游离氨基酸的全组分比较分析

倪大洲　叶文虎*　江绍慧　吕　群
黄文斌*　王　斌*　杨纯本*　胡泊新*

我们使用Beckman119CL型氨基酸分析仪生理柱的方法，对安徽大别山区的10例正常

人和10例克汀病患者的尿液进行游离氨基酸的测定。尿液里的每种氨基酸的含量以所占正常人和患者尿液中总氨基酸的百分比含量而分别予以计算。结果表明：尿液中游离氨基酸的定性鉴定，正常人与患者尿液中有23种相同的游离氨基酸。经氨基酸的含量统计分析，在正常人与患者之间差异显着（$P<0.005$）的有3种氨基酸，即苏氨酸，半胱氨酸和肌肽。

•安庆地区克汀病防治研究所，安徽

Analysis of Free Amino Acids in Urine from Normal Individuals and Cretinism

Ni Dachou Ye Wenhu Jiang Shaohuei Lu Qun
Huang Wenbing Wang Bing Yang Chunbin Hu Boxin

Free amino acids of urine from ten normal persons and ten Critinism in the region of Dabei mountains of Anhwei province were determined, using the sing -column method for physiological fluid. The instrument used was Beckman 119CL amino acid analyzer. Proportions of each amino acid to the total amount of amino acids in the urine of normal and affected individual were calculated separately. Twenty-three kinds of amino acids were identified both for the urine from normal persons and Critinism. On quantitative biometry analysis of amino acids, significant differences ($P<0.005$) were found for threonine, cysteine and carnosine between normal persons and Critinism.

1—3 人体基因组的体细胞遗传工程

1.人体和中国仓鼠杂种细胞的建立和鉴定

邱信芳 周洁民 李永全*
袁汉英 薛京伦 刘祖洞

本文以中国仓鼠肺细胞Wg3h(HGPRT⁻)和正常人的淋巴细胞作为亲本，借助仙台病毒促融，HAT培养液选择，成功地得到了FD1杂种细胞。通过细胞形态、Giemsa-11分化染色，G-分带，G6PD和LDH同功酶的电泳等一系列鉴定，以及细胞周期的研究，进一步证实杂种细胞是包含全套中国仓鼠染色体组和少数几条人的染色体（5、11、12、21、22、Y、X_q^-等的杂种，其中还存在中国仓鼠和人体染色体的易位。这一工作为人类和啮齿类杂种细胞的建立和鉴定提供了一整套方法。FD1等杂种细胞的建立将有助于进行人类基因定位以及人体基因的结构功能和表达调控的研究。

•湛江医学院

Cell Genetic Engineering of Human Genome
I. Hybrid Formation of a Chinese Hamster Cell Line and Lymphocytes and Its Identification

Qiu Xingfang Zhou Jimin Li Yongquan
Xue Jinglun Liu Zudong

Chinese hamster cell line Wg3-h(HGPRT$^-$) and normal human lymphocytes were used as parental cells for the fusion experiment in the presence of UV-inactivated Sendai virus. The resulting F1 hybrid after being selected in HAT medium retained a complete Chinese hamster genome plus a few human chromosomes (No. 5, 11, 12, 21, 22, Y,Xq$^-$.....). Besides, translocations between Wg3-h and human chromosomes were also observed. Cell morphology, Giemsa-11 staining, G-banding, glucose-6-phosphate dehydrogenase (G-6-PD) and lactate dehydrogenase (LDH) isozyme eletrophoretic analyses were used as criteria in identifying the hybrid.

This result provides a valuable information for obtaining and identifying rodent-human somatic cell hybrids and will contribute to the mapping of human genes and understanding of the structure and function of huma genome. It may also lead to answering some of the questions in gene expression and regulation as well.

1—4 人鼠杂种细胞中染色体复制周期的研究

薛京伦 邱信芳 周洁民

人体淋巴细胞的细胞周期为24小时，而中国仓鼠Wg3-h的细胞周期为12小时；人体淋巴细胞的G_1期比Wg3-h的G_1期长。杂种细胞F1是由上述两种细胞作为亲本融合而成的。我们对F1杂种细胞中人和仓鼠染色体的复制周期进行了初步研究。SCE是一种十分敏感的染色体损伤的指标，Hoechst加Giemsa染色体技术不仅简化检测SCE的方法，也为研究细胞群体分裂动力学提供了一种新工具。在杂种细胞培养液中加入每毫升最终浓度为10μg的BrdU，经24小时(Wg3-h两个细胞周期)和48小时(人体淋巴细胞两个周期)非同步化培养后收获细胞，用姐妹染色单体分化染色法(SCD)对染色体进行Giemsa分化染色；因为在DNA复制过程中，BrdU代替胸腺嘧啶掺入新合成的DNA链(M_1)，经过两个分裂周期(M_2)后，有一条染色单体的DNA双链之一含有BrdU，而另一条染色单体的DNA双链都含有BrdU；用

SCD方法染色后，可以观察到两条都被BrdU取代的DNA链不能被Giemsa染色，而呈淡染色单体；只有一条DNA链被BrdU取代的染色单体却能染成深色，在普通显微镜下较易辨别。结果发现在24小时收获的标本中人和仓鼠两种染色体都已表现出明显的染色单体分化染色，占中期细胞总数的80%；而在48小时收获的标本中，极大多数(60%)细胞的两条染色单体均已淡染，说明两条单体的DNA双链上均已掺入BrdU不能呈现分化染色，细胞已进入第三次（M_3）有丝分裂，在观察到的所有杂种细胞中未发现有仓鼠和人染色体处于不同分裂周期的情况，要末两者都有分化染色，要末两者都没有分化染色，未发现一个物种的染色体能观察到分化染色而另一个物种的染色体无分化染色。这一观察结果指出：在杂种异核体细胞中各个物种的亲本失去它们自已复制周期时间的特点，DNA合成的起点都由G_1期较短的那个种来决定。原因可能是合成DNA的复制子受多种因素的调节而启动，细胞质中可能存在这种调节因子。

Studies on the Chromosome Replication Cycles of the Human/Rodent Hybrid

Xue Jinglun Qiu Xinfang Zhou Jimin

F1 hybrid was obtained by fusing of parental cells from human lymphocytes and Chinese hamster Wg3-h cells. The cell cycle for the human Lymphocytes stimulated by PHA was 24 hrs, while Wg3-h was 12 hrs. The G_1 phase of the human lymphocytes was longer than the Chinese hamster Wg3-h cells. Sister Chromatid Exchange (SCE) is a new tool for studying the kinetics of the cell division. We added BrdU at a concentration of 10μg/ml into the F1 hybrid cultures, after incubated for 2 cell cycles, that is, 24 hrs for the Wg3-h cells and 48 hrs for the human lymphocytes. The chromosome preparations were stained with sister chromatid differentiation (SCD) staining method.

The results showed that more than 80% of the human and rodent chromosomes in the 24 hrs cultures showed SCD, more than 60% of the human and rodent chromosomes showed light staining in the 48 hrs cultures, they were all in the third mitoses. It appeared that the initiation of the DNA synthesis in the cell hybrid were determined by the parental cells which had the shorter G_1 phase.

The results suggested that the initiation of the replicon for the DNA synthesis was regulated by various factors and it seems that there is are gulator factor exists in the cytoplasm.