



華夏英才基金學術文庫

英文版

**A Profile of Genetic and  
Environmental Diseases in China**

**中国遗传及地方性疾病资源谱**

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Editor-in-chief Wei Wang (王嵬)



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北京

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# A Brief Introduction

“A Profile of Genetic and Environmental Diseases in China” is the first book describing common genetic and endemic diseases in China. Professor Wei Wang, the Chief editor, is a Chinese geneticist and epidemiologist who has been long engaged in the study of disciplines of both genetics and public health.

There are totally 13 chapters in the book. The first chapter is an introduction, which describes geographic characteristics, population size and growth, population age structure, ethnicity, mortality patterns, fertility patterns, maternal mortality, nuptiality, polygamy or monogamy, and migration. The other 12 chapters are single gene disorders, chromosomal disorders, congenital disorders, psoriasis and eczema, essential epilepsy, behavioral and psychiatric disorders, cardiovascular diseases, epidemiology and risk factors of common cancers, dietary deficiency disorders, environmental toxicity disorders, combined deficiency-toxicity disorders, and bioethical and legislative restrictions in China. A great strength is its ability to introduce the knowledge of epidemic diseases, endemic diseases, etiological factors, risk factors, and time distribution comprehensively and systematically. The book contains the most current information available on familial genetic diseases, endemic diseases, chronic diseases and bioethical and legislative restrictions in China.

The book can be used by medical students as a supplementary textbook. It is also of great value as a reference for health professionals and physicians. Multiple copies of this valuable book should also be on the shelves of all universities, medical schools and research establishments where these subjects are studied.

# **Foreword**

Genetic disorders are commonly found in every community and endemic diseases are restricted in specific geographical areas in China. Traditionally recognized hereditary disorders in the form of chromosomal, single gene and complex medical diseases have a prevalence of about 20%-25%. If acquired genetic disorders and somatic genetic disorders such as cancer are included, the prevalence is even higher. Given the huge burden of genetic disorders on patients, families and the community, it is essential to study their mechanisms and bring forth powerful preventive measures. To effectively control the genetic disorders and endemic diseases in China, the first priority is the systematic collection of basic data such as prevalence of various disorders and endemic diseases, contributing factors, and variations in expression among different ethnic groups and the general population. However, until now there has been no Chinese books or scientific journals addressing this issue.

This book collects those information mentioned above and discusses in detail, the most commonly found multigene genetic disorders, single gene genetic disorders and chromosomal genetic disorders among Chinese, and also the endemic diseases in different parts of China. The choice of the disorders in this book was based on published Chinese epidemiological data. The emphasis of this book is focused on the disease prevalence, in different populations and regions home and abroad, the trend of alterations of prevalence, and a brief introduction to the present status of research and therapy available in China and in the world. Genetic disorders and endemic diseases in minority groups in China, genetic studies in isolated and semi-isolated communities of Chinese, and major influencing factors are also discussed.

Though not an encyclopedia of genetic disorders and endemic diseases in China, this book does contain some valuable information for doctors, researchers, health workers and students.

Last but not least, I would like to thank my wife Hong Gao for her under-

standing and enduring belief in me and my two sons, Yun Wang and Patrick Wang, who are my inspiration and loves of my life.

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# CHAPTER 1

## China and its peoples

*Xiao-feng Jiang , Wei Wang*

### 1. 1 Chinese population demographics

Compared to the rest of the world, Chinese population demographics are heavily skewed to a progressively aging populations. China has 22. 9% of her people under 15 years of age and 7. 09% over 65 years of age, with a particular high proportion of aged females. In contrast, 32. 3% of the worlds population is less than 15 years of age and 6. 2% is more than 65 years of age (Table 1-1)<sup>[1]</sup>.

The proportion of the population over 65 years old varies little between different provinces, with the exception of regions with a smaller density, e. g. , Tibet, Qinghai, and Xinjiang<sup>[2]</sup>.

**Table 1-1 Composition of population by age(years)**

| Age   | Total |       |       | Male(%) |       |       | Female(%) |       |       |
|-------|-------|-------|-------|---------|-------|-------|-----------|-------|-------|
|       | 1990  | 2000  | 2004  | 1990    | 2000  | 2004  | 1990      | 2000  | 2004  |
| Group |       |       |       |         |       |       |           |       |       |
| Total | 100   | 100   | 100   | 51.45   | 51.53 | 50.85 | 48.55     | 48.47 | 49.15 |
| <15   | 27.70 | 22.90 | 19.30 | 14.39   | 12.18 | 10.41 | 13.31     | 10.72 | 8.90  |
| >65   | 5.58  | 7.09  | 8.58  | 2.55    | 3.36  | 4.11  | 3.03      | 3.75  | 4.45  |

China's rapidly aging population has already become a major problem acknowledged by the authorities, requiring the creation of special policies including health care plans.

### 1. 2 The Han and the ethnic minorities

China is a country with 56 ethnic nations distributed in different areas, all of

which have their own lifestyles. As disease patterns are affected by both “nature” (genetics) and “nurture” (environment) the study of population genetics in China is unique in scale and complexity.

The Han people are the majority. The fifth national population census in 2000 showed that there were 1159. 4 million people of Han ethnic group, accounting for 91. 6% of the total population in China. Han people live in many different areas in China, and their population is concentrated in the regions of the Yellow River, the Yangtze River, Zhu River and Songliao Plain. The Han ethnic group is an amalgamation of ancient Huaxia ethnic group with many other groups. The name Han has been used since the Han Dynasty which started in 206BCE. The most widely used spoken language is Mandarin; Despite having many spoken languages and dialects, China has had the great advantage of having only one written language, which has been a great force for unity and national identity since the writing system was formalized by Li Si and its use mandated by the Emperor Qin Shihuang in 220 BCE. Chinese characters are among the oldest characters in the world. There are more than 40, 000 words, of which 5, 000-8, 000 are frequently used. The main religions are Buddhism and Taoism. Staple foods are rice and wheat, with vegetables, meat and bean products as secondary foods. Tea and wine are traditional beverages. The marriage system is monogamy which is protected by law.

The Zhuang ethnic group is China's largest minority group. They mainly live in Guangxi Zhuang Autonomous Region, Wenshan Zhuang and Miao Autonomous State in Yunnan Province, and some of them have settled in Guangdong, Hunan, Guizhou, and Sichuan. According to the fifth census in 2000, the Zhuang population is 16, 178, 811. Most of them speak Zhuang language. Rich Zhuang people may be polygamous, while poor ones are usually monogamous. There is a strange custom persisting to this day, where a bride will continue to live in her parental home after marriage, for up to 3-5 years, the bride will return home to live in case of major festivals and harvest season came to a temporary residence husband home only moving in with her husband after having children.

The second most numerous minority is the Manchu who originated in North East China and despite their lesser numbers, ruled China during the Qing Dynasty from 1644 CE until the Republic of China was established under Sun Yat Sen in 1911 CE. For this reason, Manchu people are scattered almost all over China. Most of them live in Liaoning Province, and the rest in the provinces like Jilin, Hebei, Inner Mongolia, Xinjiang, Gansu, Shandong and also live in some big cities,

such as Beijing, Tianjin, Chengdu, Xi'an, Guangzhou, Yinchuan, etc. There are many Manchu autonomous counties such as Youyan, Fengcheng, Xinbin, Qinglong, Fengning, and several Manchu autonomous villages. According to the fifth census in 2000, the total population of Manchu has reached 10,682,263, who speak both Manchurian and Mandarin. They are monogamous. Although Manchu people have their own unique classes and culture, they are almost the same as the Han nationality now in living styles.

The Hui people are the most widely distributed minority group. The fifth census in 2000 showed their population to be 9,816,802. Hui people mainly live in Ningxia Hui Autonomous Region, and some live in Gansu, Qinghai, Henan, Hebei, Shandong and Yunnan provinces. There are several significant local communities of Hui people in China. They generally speak Mandarin, and because the majority of Hui are Muslim, they are monogamous and have food taboos, avoiding pork, animal blood and animals that die naturally.

The Lhoba ethnic minority group has the smallest population in China, scattered in the southeast part of Tibet Autonomous Region such as Luoyu, Chayu, Motuo, Milin, and Longzi counties. According to the fifth census in 2000, the population of Lhoba is 2,965. Lhoba people are generally monogamous, but polygamy prevails in some rich tribes. Traditional marriage rules are still observed, such as payment of dowry, and obligation of a man to marry his deceased brothers widow. Information about other ethnic minorities is listed in Table 1-2<sup>[1]</sup>.

**Table 1-2 Ethic groups' population, generally used language and marriage system**

| Nationality | Population (2000) | Language                                 | Marriage system                     |
|-------------|-------------------|--|-------------------------------------|
| Han         | 1,159,400,000.00  | Mandarin                                 | Monogamy                            |
| Zhuang      | 16,178,811.00     | Zhuang language                          | Most monogamy, polygamy in the rich |
| Manchu      | 10,682,263.00     | Manchu language, now generally Mandarin  | Monogamy                            |
| Hui         | 9,816,805.00      | now generally Mandarin                   | Monogamy                            |
| Miao        | 8,940,116.00      | now generally Mandarin                   | Monogamy                            |
| Uygur       | 8,399,393.00      | Uygur language, old Uygur characters     | Monogamy                            |
| Tujia       | 8,028,133.00      | now generally Mandarin                   | Most monogamy                       |
| Yi          | 7,762,286.00      | Yi language, Yi characters               | Most monogamy                       |
| Mongolian   | 5,813,947.00      | Mongolian language, Mongolian characters | Monogamy                            |

continued

| Nationality | Population (2000) | Language  | Marriage system                 |
|-------------|-------------------|---|---------------------------------|
| Tibetan     | 5,416,021.00      | Tibetan   | Most monogamy, also polygamy    |
| Bouyei      | 2,971,460.00      | Bouyei language, Chinese characters                           | Monogamy                        |
| Dong        | 2,960,293.00      | Dong language, Chinese characters                             | Monogamy                        |
| Yao         | 2,637,421.00      | Mandarin, Zhuang language                                     | Most monogamy                   |
| Korean      | 1,923,842.00      | Korean language, Korean characters                            | Monogamy                        |
| Bai         | 1,858,063.00      | Bai language, Chinese characters                              | Monogamy                        |
| Hani        | 1,439,673.00      | Hani language   | Monogamy                        |
| Kazak       | 1,250,458.00      | Kazak language  | Monogamy                        |
| Li          | 1,247,814.00      | Li language   | Most monogamy                   |
| Dai         | 1,158,989.00      | Dai language  | Most monogamy, Headman polygamy |
| She         | 709,592.00        | She language, now generally Chinese characters                | Most monogamy                   |
| Lisu        | 634,912.00        | Lisu language   | Monogamy                        |
| Gelao       | 579,357.00        | Now generally Mandarin  | Monogamy                        |
| Dongxiang   | 513,805.00        | Dongxiang language, now generally Chinese characters          | Monogamy                        |
| Gaoshan     | 400,000.00 or so  | Now generally Chinese characters                              | Monogamy                        |
| Lahu        | 453,705.00        | Lahu language, Mandarin, Dai language                         | Monogamy                        |
| Shui        | 406,902.00        | Shui language, now generally Chinese characters               | Monogamy                        |
| Va          | 396,610.00        | Va language   | Monogamy                        |
| Naxi        | 308,839.00        | Naxi language   | Monogamy                        |
| Qiang       | 306,072.00        | Qiang language, now generally Chinese characters              | Most monogamy                   |
| Tu          | 241,198.00        | Tu language, now generally Chinese characters                 | Most monogamy                   |
| Mulam       | 207,352.00        | Mulam language, Maonan language, Shui language, Dong language | Most monogamy                   |
| Xibe        | 188,824.00        | Xibe language   | Most monogamy                   |
| Kirgiz      | 160,823.00        | Kirgiz language   | most monogamy, some polygamy    |
| Daur        | 132,394.00        | Daur language, now generally Chinese characters               | Monogamy                        |
| Jingpo      | 132,143.00        | Jingpo language   | Most monogamy, some polygamy    |