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The Survival and Medical Dilemma of Rare Disease Patients in the Greater Bay Area

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Abstract:

The population of rare disease patients in China faces many difficulties in disease diagnosis and treatment, medical security, economic burden, quality of life, social assistance, employment opportunities, mental health, and other aspects. It is of great significance to identify the common problems faced by this group and try to propose possible solutions, calling on all sectors of society to pay attention to the rare disease minority group, help them solve their survival and security difficulties, and enable more patients to return to society. This paper investigates and addresses the survival and security challenges faced by rare disease minority patients in the Guangdong-Hong Kong-Macao Greater Bay Area (GBA) of China. Through a survey of 223 rare disease patients in the Greater Bay Area, the study reveals that these patients face many challenges in diagnosis, treatment, drug access, and economic support. The conclusion emphasizes the necessity of improving diagnostic efficiency, accelerating drug development, and improving the medical insurance system. Thus, it further puts forward practical suggestions.

Keywords: GBA; rare disease; disease burden; survival dilemma.

1. Introduction

Rare diseases are a type of disease with extremely low incidence and rare occurrence, usually chronic and severe, often endangering life. Different countries and regions define rare diseases according to different standards. The Orphan Drug Act of 1983 in the United States defined rare diseases as those with a population of less than 200,000 patients (approximately 0.6 ‰) in the country. China mainly defines rare diseases through disease catalogs. In 2018, the National Health Commission of China, together with five departments, released the "First Batch of Rare Disease Catalog", which included 121 diseases in the scope of rare diseases. In 2023, the "Second Batch of Rare Disease Catalog" was released, which included a total of 86 rare diseases.

It is estimated that there are approximately 20 million rare disease patients in China [1]. The diagnosis and treatment of rare disease patients are international challenges. According to the 2019 China Rare Disease Comprehensive Social Survey Report, 42% of patients worldwide reported having been misdiagnosed, and over 40% of patients were diagnosed for more than a year. In terms of treatment drugs, less than 10% of known rare diseases worldwide have drugs or treatment plans, with less than 5% of drugs introduced to China. Moreover, due to the high cost of drugs, most patients' families find it difficult to afford the expenses.

Due to different policies in various regions, in areas lacking national medical insurance payment and commercial health security policies, most patients still cannot afford high medical expenses. They face many difficulties in terms of economic burden, disease diagnosis and treatment, employment opportunities, psychological pressure, and quality of life.

The Greater Bay Area is located in southern China and serves as the forefront of China's economic development. It has abundant medical resources and an advanced medical service system, providing high standard medical security for regional residents and gradually becoming an important medical and health service center in the Asia Pacific region [2]. Studying and addressing the survival and security challenges faced by rare disease minority patients in the Guangdong Hong Kong Macao Greater Bay Area can help pave the way and provide examples for related issues nationwide and even globally.

2. Research Methods and Sample Information

This study applies a combination of qualitative and quantitative analysis methods to investigate the survival and security issues of patients with major rare diseases in the

Greater Bay Area. The author conducted a survey of 223 patients with more than 10 rare diseases, including SMA, Gaucher's disease, Pompe's disease, Fabre's disease, MPSI, MPSII type, MS, and juvenile rickets, through a questionnaire survey. In addition, a total of 9 patients underwent telephone interviews, and four medical insurance experts participated in in-depth interviews.

Among these 223 patients, most of them are over 46 years old (85.65%). There are 148 males (66.37%) and 75 females (33.63%), 17 unmarried (7.62%), 192 married (86.1%), 10 divorce (4.48%) and 4 widowed (1.79%). About their identity of insurance, 105 of them are urban and rural residents (47.09%), 106 are urban employees (47.53%), and 12 are uninsured (5.38%). Twenty-nine people have illiterate or primary school education (13%), 63 people have junior high school education (28.25%), 59 people have junior high school, polytechnic or vocational school education (26.46%), 65 people have undergraduate or college education (29.15%), and seven people have master's degree or above (3.14%).

Patient's age, occupation, marital status, and annual family income and some other information are as follows (Table 1):

Table 1. Patients' information

	N	%
occupation		
unemployed	56	25.11
Civil servants or personnel of public institutions	25	11.21
Professional technical personnel	11	4.93
Enterprise employees	23	10.31
Part time or freelance work	10	4.48
farmer	30	13.45
worker	12	5.38
student	1	0.45
retired personnel	51	22.87
other	4	1.79
annual household income (CNY)		
below 30k	85	38.12
30k~60k	46	20.63
60k~90k	29	13
90k~120k	26	11.66
120k~150k	11	4.93
150k~180k	6	2.69
180k~210k	7	3.14
210k~240k	5	2.24
240k~270k	0	0
270k~300k	3	1.35
300k~330k	2	0.9
330k~360k	0	0
360k~390k	0	0
390k~420k	0	0
420k~450k	1	0.45
over 450k	2	0.9

family members living together		
live alone	20	8.97
with parents	41	18.83
with spouse	161	74.44
with kids	81	36.77
With relatives	2	0.9
The main source of daily expenses		
work income	58	26.01
pension	72	32.29
income of family members	68	30.49
financial assistance from relatives and friends	13	5.83
social donations	0	0
government guarantee	7	3.14
other	5	2.24

3. Patient Treatment Status

The data shows that 81.86% of patients are dissatisfied with the current treatment effect, including 47.98% very dissatisfied and 33.63% dissatisfied. To understand the reasons behind this phenomenon, the author conducted research through the following aspects.

In terms of diagnosis, the data suggests that 41.7% of patients are diagnosed within 1-3 years, 25.56% within 3-5 years, 25.56% within five years or more, and only 7.17% of patients are diagnosed within one year.

Moreover, the above data demonstrates that the current treatment methods for patients mainly include drug therapy (90.58%), physical therapy (24.22%), and psychological support (19.73%). Among them, drug therapy is the most important treatment method, accounting for the highest proportion; Physical therapy and psychological support come second. There are relatively few options for other treatment methods, accounting for 7.62%.

It is also worth noting that 68.61% of patients have a negative evaluation of drug accessibility, with 49.78% rated as poor and 18.83% rated as poor (Fig. 1). In addition, 75.34% of patients believe that the lack of effective treatment plans is the biggest obstacle during the treatment process (Fig. 2). This may indicate that patients face the significant dilemma of having no available treatment options while also struggling to find a single effective drug.

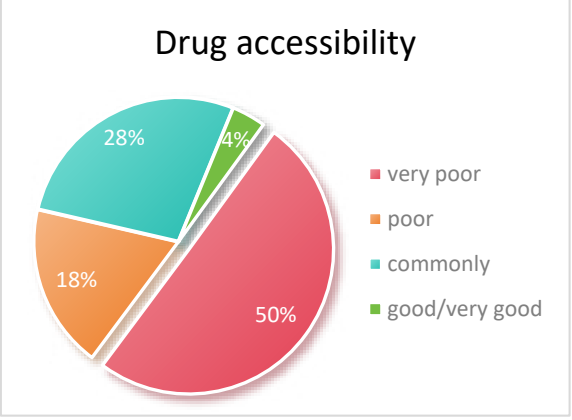

Fig. 1 Drug accessibility

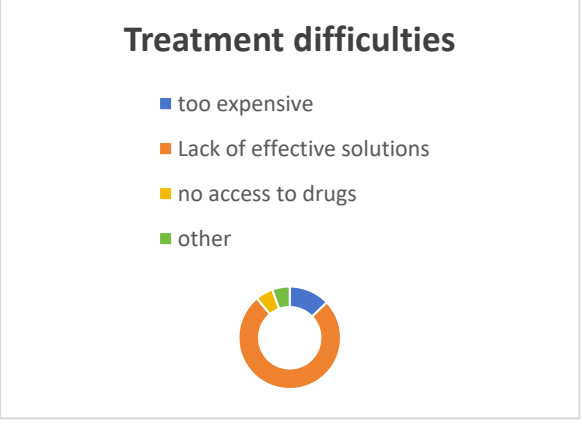

Fig. 2 Difficulties of treatment

4. Patient Survival Status

Among 223 rare disease patients, a significant 74.89% consider their quality of life to be poor, with 28.25% rating it as poor and 46.64% as very poor. Approximately 21% consider their quality of life to be average, while only 4.04% rate it as good or very good (Fig. 3).

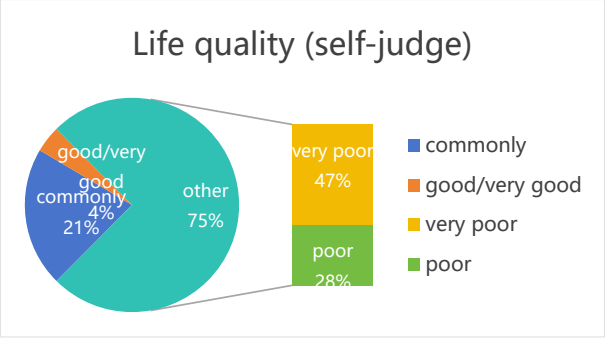

Fig. 3 Life quality of patience

In order to explore variables that are correlated with patients' quality of life, the author conducted SPSS correlation analysis on patients' quality of life, diagnosis time, drug affordability, drug availability, and impact on career development. Table 2 shows the specific data of the above variables:

Table 2. SPSS analysis

	N	%
life quality of patience		
good/very good	9	4.04
commonly	47	21.08
poor	63	28.25
very poor	104	46.64
diagnosis time		
In 1 year	16	7.17
1~3 year	93	41.7
3~5 year	57	25.56
Over 5 years	57	25.56
Drug affordability		
really hard	104	46.64
hard	63	28.25
commonly	51	22.87
easy	5	2.24
very easy	0	0
the availability of drugs		
very poor	111	49.78
poor	42	18.83

commonly	61	27.35
good	8	3.59
very good	1	0.45
The impact of illness on career development		
none(1 point)	10	4.48
not too big(2 point)	6	2.69
significant impact(3 point)	21	9.42
Serious impact (4 point)	24	11.21
can't work(5 point)	158	72.2

After the above data was analyzed using SPSS, the results are shown in Table 3 below:

Table 3.

subject	average	standard deviation	Life quality	Drug availability	Diagnose time	impact of illness on career development	Drug affordability
Life quality	4.11	1.02	1				
Drug availability	1.79	0.96	-0.40**	1			
Diagnose time	2.69	1.00	-0.15	0.08	1		
impact of illness on career development	4.48	1.03	0.46**	-0.11	-0.26**	1	
Drug affordability	1.82	0.89	-0.22*	0.34**	0.03	-0.16	1

(* $p<0.05$ ** $p<0.01$)

Based on the above SPSS analysis, the following conclusions can be drawn:

a) Quality of life: The average value of patients' evaluation of their current quality of life is 4.11, with a standard deviation of 1.02, indicating that most patients have a poor evaluation of their quality of life.

b) The patient's evaluation of quality of life is negatively correlated with the availability of therapeutic drugs (-0.40 * *), indicating that the poorer the drug availability, the lower the patient's evaluation of quality of life.

c) Diagnosis time: The average value is 2.69 years, with a standard deviation of 1.00 years, and there is no significant correlation with quality of life and availability of treatment drugs.

d) Drug accessibility and drug cost burden: The degree of drug accessibility is positively correlated with the affordability of drug costs (0.34 * *), and negatively correlated with quality of life (-0.22 *), meaning that the more difficult it is to afford drug costs, the worse the accessibility of drugs

e) The affordability of drug costs: with an average value of 1.82 and a standard deviation of 0.89, it is significantly positively correlated with the availability of therapeutic drugs (0.34 * *) and negatively correlated with quality of life (-0.22 *), indicating that higher costs may lead to poorer drug availability and potentially lower quality of life.

f) Occupational impact and drug cost burden: The degree of impact of diseases on employment and career development is negatively correlated with the affordability of drug costs (-0.16), indicating that the more difficult it is to afford drug costs, the greater the impact on the profession.

The author found that economic factors greatly affect the quality of life for patients, often leading to severe survival difficulties. Survey data shows that about 75.34% of patients believe that the cost of disease treatment is the main factor leading to their economic burden. It is worth noting that 38.12% of the 223 patients had an annual household income of less than 30,000 yuan, while 20.63% reported a median income between 30,000 and 60,000 yuan. When excluding all reimbursements and assistance, the proportion of total personal expenses paid by patients in the past year exceeding 100,000 yuan is 23.77%, 50,000 to 100,000 yuan is 25.56%, and 30,000 to 50,000 yuan is

25.11%. This means that the annual treatment cost for patients can be twice or even more than the total household income.

The disease has already seriously affected patient's career and ability to work, leading to reduced income, and the difficulty in affording medication has further increased the burden on rare disease patients and their families. Like a set of dominoes, the fall of one ring will inevitably affect another ring. Unfortunately, for patients with rare diseases, this process is constantly deteriorating.

Furthermore, when patients are asked if they have negative feelings (such as low mood, despair, anxiety, and depression), 33.18% always have them, 35.87% often have them, and 22.87% sometimes have them. In addition, 34.98% of patients stated that they have no confidence in facing the future, while only 2.69% expressed confidence. It has been proven that rare disease patients also face serious psychological pressure, aside from physical ailments.

The author has observed the despair and demands of some patients in qualitative research, which can be roughly summarized into the following four points: First, "the existing drugs are too expensive, and I hope they can be reduced in price"; Second, "some diseases are incurable in China, and we hope to introduce or launch generic drugs"; Third, looking forward to the emergence of new drugs to enable more rare patients to receive treatment; Lastly, "I hope society can increase its attention to rare diseases and strengthen the protection of rare disease patients".

5. Discussion

Although Chinese society has paid increasing attention to the issue of rare diseases in recent years, and the Chinese government has introduced multiple policies to gradually solve the difficulties faced by the rare disease population in terms of diagnosis, treatment, medication accessibility, and affordability, significant obstacles remain in the diagnosis, treatment, and protection of rare diseases patients.

Data from this GBA patient survey shows that only 7.17% of patients are diagnosed within one year, 41.7% of patients are diagnosed within 1-3 years, and 25.56% of patients are diagnosed with rare diseases for more than five years. According to the 2020 China Rare Disease Comprehensive Social Survey, nearly 70% of healthcare workers out of 38,634 reported that they were not familiar with rare diseases. In a survey of 20,804 patients with 33 rare diseases, 42% of patients reported that it takes an average of 4.26 years to receive an accurate diagnosis after initially being misdiagnosed [3].

The development of drugs for rare disease faces significant challenges, including high cost, difficulties in conducting clinical trials, and limited number of companies developing rare disease drugs. As a result, Chinese patients have long faced the dilemma of 'having drugs overseas but not domestically'. Although in recent years, the GBA has been able to directly introduce rare disease drugs that have already been marketed overseas, some patients still face the dilemma of having no drugs available.

Moreover, due to issues such as limited drug accessibility and high treatment costs, the proportion of rare disease receiving treatments remain low. According to a survey conducted by the China Rare Disease Alliance on 20804 rare disease patients, nearly one-third (26.1%) of the patients had not received treatment or had never received treatment. Among this group of patients, about half of them gave up treatment due to the high cost of medical expenses that they could not afford [3].

The Chinese government is still exploring and improving the medical security model for rare diseases. Among the existing 207 rare disease catalogs in China, only 90 diseases have corresponding drugs included in the national medical insurance drug catalog. At the local level, some provinces provide certain protection benefits for a small number of high-value rare disease self-funded drugs through special guarantee funds. In 2019, Zhejiang Province established the first provincial-level special fund for the guarantee of rare disease medication in China, prioritizing the inclusion of two rare disease treatment drugs, including Gaucher and phenylketonuria, in the scope of medication reimbursement [4]. In 2020, Foshan City, Guangdong Province issued the "Notice of the Office of the People's Government of Foshan City on Issuing the Medical Assistance Measures of Foshan City", which includes diseases in the national rare disease catalog in the scope of medical assistance [5].

Through interviews with four medical insurance policy experts, the author stated that universal commercial supplementary health insurance has gradually become an important force in providing multi-level protection for rare diseases. The core cities of GBA, Guangzhou and Shenzhen, both provide government led commercial supplementary health insurance to cover self-funded drugs outside the basic medical insurance list. Rare disease patients in Guangzhou can be reimbursed up to 70% annually and up to 1 million yuan annually after purchasing Guangzhou's commercial health insurance "Sui Sui Kang" [6]. Rare disease patients in Shenzhen can also be reimbursed 70% after purchasing Shenzhen commercial insurance, with a maximum annual reimbursement of 500000 yuan.

Although some patients have received certain cost compensation through basic medical insurance, commercial insurance, medical assistance, etc., for most patients, the treatment drugs for rare diseases are expensive and the cost paid is not enough to cover the total treatment cost.

Moreover, policies in some cities are inconsistent, and there are still patients in some cities who cannot afford high treatment costs due to the lack of commercial insurance or medical assistance policies and can only give up treatment. In this GBA survey, when asked if you can reimburse treatment expenses through the national basic medical insurance, the data showed that only 0.46% of patients can be fully reimbursed for treatment expenses, 63.93% of patients can only be partially reimbursed for treatment expenses, and up to 35.62% of patients cannot be reimbursed for all treatment expenses. Over half of cities does not have a commercial insurance policy, and 12.33% of patients stated that they can be partially reimbursed through commercial insurance, but the reimbursement rate is below 50%.

Although GBA is a relatively developed region in China's economy, rare disease patients still face multiple challenges due to the lack of comprehensive and systematic institutional design for medical insurance for rare diseases in China, the absence of protection for "high-value rare disease drugs", and significant differences in protection benefits between different regions. Difficulties in disease diagnosis, insufficient treatment drugs, and inadequate medical security have led to enormous economic and psychological pressure on patients and their families, seriously affecting their quality of life and social development opportunities.

Rare diseases are both a critical livelihood issue and a matter of social civilization. The protection of the basic health rights of patients with rare diseases fundamentally reflects the level of civilization of a country or region. The economic development level of the GBA provides a solid economic foundation for high drug reimbursement, and its strong industrial foundation and financial strength can support high medical expenses. In the future, the GBA should continue to strive to explore a rare disease diagnosis, treatment, and protection model that is in line with GBA's characteristics. This effort should focus on addressing the survival and protection difficulties faced by rare disease patients in the GBA, with the goal of benefiting a larger number of patients.

6. Conclusion

The sample size of patients in this survey is moderate, and a multidimensional investigation was conducted on the GBA rare disease population. At the same time, valuable data and results were obtained by combining policy research, expert interviews, literature analysis, and other methods. Based on the research results and the level of economic development in GBA, the author proposes the following suggestions.

First is to improve diagnostic efficiency and accuracy. This requires establishing a rare disease diagnosis center to ensure that patients can receive accurate diagnosis. For example, referring to the Rare Disease Diagnosis Center (CDMR) model in France, this model significantly improves the speed and accuracy of rare disease diagnosis by integrating resources and professional knowledge. The implementation of a rare disease education and training program can also enhance the awareness and diagnostic abilities of doctors for rare diseases by regularly holding professional training and seminars.

Second, it is important to accelerate the development and market launch of drugs for rare diseases by establishing special funds to support research, encouraging research institutions and companies to cooperate in tackling key issues, and accelerating the development of new drugs. For example, providing funding subsidies and tax incentives during the research and development phase to reduce research and development costs and corporate risks. Moreover, introducing incentive measures like the Orphan Drug Act in the United States can provide preferential policies such as market exclusivity for companies and encourage them to invest in the research and development of rare disease drugs.

Third, local governments should improve the medical insurance system for rare diseases. The system should cover more rare disease drug costs, especially those newly launched drugs that are expensive but have significant therapeutic effects. At the same time, they can establish a special fund to provide protection for high-value drugs that are not included in national medical insurance.

Furthermore, local governments should strengthen the balanced allocation of regional medical resources by increasing investment in grassroots medical institutions and improving their equipment and doctor skills, especially in economically underdeveloped areas. For example, investing in primary healthcare equipment through financial subsidies or public-private partnership (PPP) models to improve the quality of medical services. Moreover, they can promote cooperation and resource sharing between superior and subordinate medical institutions. They can learn from the experiences of Beijing and Shanghai in achieving a high proportion of local confirmed cases and improving the diagnosis and treatment capabilities in remote areas through technologies such as telemedicine.

Lastly, local institutions can establish a GBA rare disease patient organization to provide psychological support and social assistance for patients with rare diseases. These organization can carry out vocational skills training and social integration projects for rare disease patients to help them reintegrate into society and realize their self-worth. This can refer to successful cases abroad, such as

the “Work Redesign” program in the United States, which helps people with disabilities adapt to the work environment and increase employment rates.

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