

# An update on China's national policies regarding rare diseases

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**SUMMARY** Over the past few years, China has paid greater attention to the topic of rare diseases. The Chinese Government has made considerable efforts to gradually improve the situation of patients with rare diseases in terms of diagnosis and treatment, access to medication, and affordability of care. The National Health Commission implemented a raft of measures, including the first Catalog of Rare Diseases, establishment of a rare diseases alliance, establishment of the Network for Collaboration in Rare Disease Diagnosis and Treatment, formulation of the Guidelines for the Diagnosis and Treatment of Rare Diseases, sharing of diagnostic and treatment information, and creation of expert committees, to ensure the standardization of rare disease diagnosis and treatment and to promote the improvement of rare disease diagnosis and treatment capabilities nationwide. In order to encourage the research, development, and production of drugs to treat rare diseases, the National Medical Products Administration has drafted a series of policies to accelerate the review and approval of drug registration and to support the research and development of drugs to treat rare diseases. The Ministry of Finance has also helped to implement tax incentives for drugs to treat rare diseases, to encourage the marketing and importation of drugs to treat rare diseases, and to reduce the cost of drugs for patients with rare diseases. Through adjustment of the List of Drugs Covered by National Medical Insurance, the National Healthcare Security Administration has covered an increasing number of drugs to treat rare diseases under basic medical insurance. It has also negotiated to reduce the price of some drugs to treat rare diseases, further reducing the economic burden on patients with rare diseases.

**Keywords** rare diseases, drugs, policies, China

## 1. Introduction

Rare diseases refer to diseases with a very low prevalence. The definition of rare diseases varies in different countries around the world. An article in the internationally renowned academic journal *Nature Reviews Drug Discovery* in 2020 stated that by integrating multiple rare disease-related knowledge bases and databases, more than 10,000 rare diseases have been evaluated (1). Currently, China has more than 20 million people with rare diseases who have very few treatment options (2). Misdiagnosis and difficulties related to drug options are among the main challenges in diagnosing and treating rare diseases. China has emphasized the diagnosis, treatment, and relief of rare diseases. Since China implemented the New Drug Registration Regulation in May 1999, it has made considerable efforts to support the development of rare diseases. The current study describes three key aspects of recent national policies regarding rare diseases in

order to examine the current state of management of rare diseases in China.

## 2. Three key aspects of national policies on rare diseases

### 2.1. Promoting the improvement of rare disease diagnosis and treatment capabilities nationwide

We listed current national policies on diagnosis and treatment of rare diseases in Table 1. In May 2018, five authorities announced the "First Catalog of Rare Diseases," which included 121 rare diseases (3). This was the first time that the Chinese Government drafted a list of rare diseases, providing an opportunity to discuss rare diseases in public.

In October 2018, a Chinese medical research alliance was set up to accelerate the study of rare diseases (4). The China Alliance for Rare Diseases consists of more than 50 entities including medical

**Table 1. Current national policies on diagnosis and treatment of rare diseases**

Effective time ( <i>Ref.</i> )	Issuing agency	Policy & Regulation	Content about rare diseases
May, 2018 (3)	Five authorities	China's First National List of Rare Diseases	121 rare diseases
October, 2018 (4)	China Alliance for Rare Diseases	An explanation of China's first Catalog of Rare Diseases	It includes the basic concepts, clinical manifestations, diagnosis, differential diagnoses, and treatment principles for 60 rare diseases.
February, 2019 (5)	National Health Commission	Notice on establishing the National Network for Collaboration in Rare Disease Diagnosis and Treatment	To establish a mechanism to facilitate collaboration, to relatively centralize diagnosis and treatment, and to provide two-way referrals for patients with rare diseases.
February, 2019 (6)	National Health Commission	Guidelines for the Diagnosis and Treatment of Rare Diseases (2019 ed.)	The definitions, causes, and procedures for diagnosis and treatment of 121 rare diseases.
October, 2019 (7)	National Health Commission	Notice on entry of information on the diagnosis and treatment of rare diseases	To establish a registry for patients with rare diseases.
January, 2020 (8)	National Health Commission	Office of the National Network for Collaboration in Rare Disease Diagnosis and Treatment	The Office is mainly responsible for daily contact with and management of hospitals in the collaborative network.
September, 2020 (9)	National Health Commission	Notice on publication and dissemination of the roster of the National Health Commission's second Expert Committee on the diagnosis and treatment of rare diseases and welfare for patients	The special committee will make recommendations to relevant departments.

facilities, universities, academic institutions, and companies. The Alliance is instrumental in concentrating resources and will encourage the treatment of rare diseases in China. During its launch on October 24, the Alliance published an explanation of China's first Catalog of Rare Diseases.

In February 2019, the National Health Commission issued the "Notice on Establishing the National Network for Collaboration in Rare Disease Diagnosis and Treatment," and 324 hospitals were included in the collaborative network. Member hospitals are required to include drugs to treat rare diseases in the hospital formulary and essential drug catalog in a timely manner, to conduct clinical monitoring of drugs to treat rare diseases, to warn of shortages and to report information as required, and to strive to meet the clinical demand for drugs (5). Enhanced coordination with the catalog of drugs to treat rare diseases at member hospitals, inter-hospital adjustment of drugs, and enhanced distribution and logistics allow patients with rare diseases to more easily obtain medicines nearby. The establishment of the National Network for Collaboration in Rare Disease Diagnosis and Treatment marks the beginning of the establishment of a mechanism for collaboration in the diagnosis and treatment of rare diseases and also provides a reference for treatment paths for patients with rare diseases in various regions.

In February 2019, China's first Guidelines for the Diagnosis and Treatment of Rare Diseases were published (6). Experts referred to relevant international guidelines while drafting the Chinese guidelines. The guidelines include definitions of 121 rare diseases included in the catalog released by the National Health

Commission, their causes, and procedures for diagnosis and treatment in order to improve the diagnosis and treatment of rare diseases and to benefit patients in China. In general, medical facilities in China lack personnel with sufficient skills to effectively diagnose and treat rare diseases, and the guidelines can help to train medical staff and improve their diagnostic and treatment skills.

There is no official figure on the number of people with rare diseases in China. In October 2019, the National Health Commission announced a notice on entry of information on the diagnosis and treatment of rare diseases in a bid to establish a system to collect relevant data to understand the epidemiology, clinical diagnosis and treatment of, and welfare for rare diseases in China. Now that data are being collected, the diagnosis and treatment of rare diseases and access to medicines should improve (7).

In January 2020, the National Health Commission established an Office for the National Network for Collaboration in Rare Disease Diagnosis and Treatment in order to enhance the organization and management of the National Network for Rare Disease Diagnosis and Treatment, to facilitate collaboration, and to effectively fulfil the overall role of a collaborative network (8).

Building on the original expert committee, the roster of the second expert committee on the diagnosis and treatment of rare diseases and welfare for patients was updated in September 2020 (9). Under the leadership of the National Health Commission, the special committee will research and define rare diseases and offer recommendations for changes to the rare disease catalog in conjunction with conditions in China,

**Table 2. Current national policies on drugs to treat rare diseases**

Effective time (Ref.)	Issuing agency	Policy/Regulation	Content related to rare diseases
October, 2017 (10)	The General Office of the State Council	The Opinions of the State Council on further reform of the review and approval system to encourage innovations in pharmaceuticals and medical devices.	To support the development of drugs and medical devices to treat rare diseases.
April, 2018 (11)	The State Council	Opinions on reforming and improving policies to ensure the supply and govern the use of generic drugs	To encourage the creation of generic drugs to treat rare diseases.
October, 2018 (12)	National Medical Products Administration	Guidelines for the Registration and Review of Medical Devices to Prevent and Treatment Rare Diseases	To resolve difficulties with the clinical evaluation of medical devices used to prevent and treat rare diseases.
March, 2019 (13)	Ministry of Finance of the People's Republic of China	Notice on the value-added tax policy for drugs to treat rare diseases	The VAT rate for imported drugs to treat rare diseases has been reduced to 3%.
August, 2019 (14)	Standing Committee of the National People's Congress	The Pharmaceutical Administration Law	Greenlights the development and manufacture of drugs to treat rare diseases.
October, 2019 (15)	The Central Committee of the Communist Party of China and the State Council	The Opinions on promoting the passing down of, innovation in, and the development of traditional Chinese medicine	Encourages the use of traditional Chinese medicine to treat rare diseases.

draft and update technical specifications and devise clinical pathways for the prevention and treatment of rare diseases, screen for rare diseases, and make recommendations to relevant departments regarding diagnosis and treatment, medications, rehabilitation, and welfare for patients.

## 2.2. Encouraging the research, development, and production of drugs to treat diseases

Current national policies on drugs to treat rare diseases were summarized in Table 2. In October 2017, the General Office of the Central Committee of the Communist Party of China and the General Office of the State Council issued its "Opinions of the State Council on further reform of the review and approval system to encourage innovations in pharmaceuticals and medical devices" to encourage and accelerate the development of drugs and medical devices for rare diseases (10).

China is promoting research on and increased availability of generic drugs with improved quality and efficacy in order to lower healthcare costs and to better meet public demand, according to a document issued by the State Council on April 3, 2018 (11). Drug companies are encouraged to make generic versions of drugs that are essential for clinical treatment and that are in short supply, and especially drugs to treat major infectious diseases, rare diseases, pediatric diseases, and public healthcare crises.

In October 2018, the State Drug Administration drafted the Guidelines for the Registration and Review of Medical Devices to Prevent and Treatment Rare

Diseases in order to implement the "Opinions on further reform of the review and approval system to encourage innovations in pharmaceuticals and medical devices" from the General Office of the Central Committee of the Communist Party of China and the General Office of the State Council (Notice No. 42 [2017]) (12). These guidelines focus on benefiting patients with rare diseases by scientifically resolving difficulties with the clinical evaluation of medical devices to prevent and treat rare diseases, reasonably reducing the clinical use of those devices, and promoting the clinical introduction of those devices as soon as possible through conditional approval.

A total of 21 drugs to treat rare diseases and 4 active pharmaceutical ingredients were the subject of China's first attempt at a policy to reduce the value-added tax (VAT) on certain drugs, which took effect in March 2019. The policy aimed to lower the cost for patients with rare diseases and to encourage drug development by the pharmaceutical industry. According to the policy, the VAT rate for imported drugs to treat rare diseases was reduced to 3%. The VAT for the production and sale of drugs to treat rare diseases was also reduced to 3% as of March 1 (13).

In August 2019, the Pharmaceutical Administration Law of the People's Republic of China encouraged innovation in the development of drugs that have confirmed or special curative effects or a new mechanism of action and drugs that can cure life-threatening or rare diseases. The Law greenlights urgently needed drugs, new drugs, drugs to treat pediatric diseases, and drugs that could prevent or cure serious infectious or rare diseases (14).

**Table 3. Current national policies on medications for patients with rare diseases**

Effective time (Ref.)	Issuing agency	Policy/Regulation	Content related to rare diseases
August 2019 (16)	National Healthcare Security Administration and Ministry of Human Resources and Social Security	List of drugs for national basic medical insurance, workmen's compensation, and maternity insurance.	New drugs to treat rare diseases are added.
January 2020 (17)	National Healthcare Security Administration	China implements a new list of drugs covered by national medical insurance.	7 drugs to treat rare diseases are included in the List of Drugs Covered by National Medical Insurance.
March 2020 (18)	National Health Commission	Opinions on further reform of the medical insurance system.	Cites the need to explore forms of welfare to cover drugs to treat rare diseases.
January 2021 (19)	Ministry of Finance	Import tariffs on some commodities are adjusted.	No tariffs are imposed on constituents of drugs to treat rare diseases.
December 2020 (20)	National Healthcare Security Administration and Ministry of Human Resources and Social Security	Notice on the drug list for national basic medical insurance, workmen's compensation, and maternity insurance (2020 ed.).	Some drugs to treat rare diseases have been added to the latest national essential drug list.
January 2020 (21)	The State Council of the People's Republic of China	Further reform of centralized bulk drug purchasing to ease the financial burden on patients.	Special arrangements will be made to procure drugs to treat rare diseases.

In October 2019, the Opinions of the Central Committee of the Communist Party of China and the State Council on promoting the passing down of, innovation in, and the development of traditional Chinese medicine stated that under the framework of the central financial science and technology plan (special projects, funds, *etc.*), China will conduct clinical research on the prevention and treatment of major, refractory, and rare diseases and emerging infectious diseases and accelerate the research and development of new traditional Chinese medicines (15).

### 2.3. Improving access to medications for patients with rare diseases

As shown in Table 3, some policies on medications for patients with rare diseases were implemented since 2019. In August 2019, a total of 148 new drugs were added to the regularly referenced portion of the drug list for national basic medical insurance, workmen's compensation, and maternity insurance. The added drugs include national essential drugs, drugs to treat major diseases such as cancer and rare diseases, drugs to treat chronic diseases, and drugs to treat pediatric diseases. In addition, some of the aforementioned types of drugs, and especially those to treat cancer and rare diseases, are mainly included in the list to be negotiated. If reasonable prices are negotiated in the next step, then those drugs will be included in the list as required (16).

China's new list of drugs covered by national medical insurance came into effect on January 1, 2020. It includes 70 new drugs with prices reduced by 60.7%, on average. Some 22 anti-cancer drugs, 7 drugs to treat diseases, 14 drugs to treat chronic diseases, and 4 drugs to treat

pediatric diseases will be included in the list. After the price reduction and medical insurance reimbursements, the financial burden on patients should be eased by more than 80% (17).

In March 2020, the "Opinions of the Central Committee of the Communist Party of China and the State Council on further reform of the medical insurance system" clearly stated that "forms of welfare to cover drugs to treat rare diseases need to be explored" (18). This shows that the Chinese Government is seeking to improve welfare for patients with rare diseases.

In order to reduce the economic burden on patients and improve the quality of life of the people in 2021, a notice issued by the Ministry of Finance in December 2020 stipulated that no tariffs would be imposed on the second set of anti-cancer drugs, constituents of drugs to treat rare diseases, and foods needed by particular patients (19).

After considerable reductions in their prices, a total of 119 drugs have been added to the latest national essential drug list for reimbursement, according to the National Healthcare Security Administration on December 28, 2020. This long-expected move will greatly relieve the financial burden on patients with serious conditions ranging from cancer and rare diseases to COVID-19 (20).

China will proceed with its centralized bulk drug purchasing program and regularly seek to lower medical costs for the general public, as decided at the executive meeting of the State Council chaired by Premier Li Keqiang on January 15, 2021. Special arrangements will be made to procure drugs to treat rare diseases (21).

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