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How to evaluate rare disease policy effectiveness based on policy modeling consistency (PMC) index model: A quantitative assessment of policy implementation in China

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SUMMARY: Based on the Policy Modeling Consistency (PMC) index model, this study systematically evaluates 19 national-level and 13 regional-level rare disease policy documents from 2016 to 2025, aiming to reveal the structural characteristics and consistency level of China's rare disease policy system. Using ROSTCM 6.0 software, high-frequency keywords and semantic networks from the policy texts were extracted to identify policy focus and thematic evolution. A PMC evaluation system was constructed to assign values and conduct a visual analysis of the sample policies. Results show that the average PMC index for China's rare disease policies is 5.31 (national level) and 4.94 (local level), falling within the "excellent–acceptable" range overall, indicating that the structure of China's rare disease policy system is well-developed. Among the indicators, policy nature (X2), policy function (X8), and outcome orientation (X4) scored higher, reflecting strategic planning and institutional characteristics at the national level. In contrast, incentive measures (X5), patient support (X6), and social support (X9) scored lower, highlighting deficiencies in research and development incentives, social integration, and long-term support mechanisms. Further comparisons reveal that national policies emphasize top-level design and institutional development. In contrast, local policies focus more on exploratory efforts in medical assistance and policy innovation. To optimize China's rare disease policy system, it is essential to improve incentive and social support systems, promote coordinated legislation between national and local levels, establish a national rare disease information platform and a dedicated fund, and build a comprehensive policy chain.

Keywords: policy evaluation, PMC index, rare diseases, policy evaluation

1. Introduction

Rare diseases are conditions with extremely low prevalence and small patient populations, most of which are chronic, severe, or life-threatening. No unified international definition exists, with classification typically based on prevalence or patient numbers (*1*). Over 10,000 rare diseases have been identified to date, accounting for approximately one-tenth of all human diseases (*2*). By the end of 2024, the global rare disease patient population had exceeded 300 million, with a trend toward earlier onset and longer disease duration (*3*). The Chinese Medical Association has proposed reference criteria for rare diseases: conditions with a prevalence below 1/500,000 or a neonatal incidence rate below 1/10,000 are classified as rare diseases (*4*).

To address rare disease challenges, multiple countries have established systematic policy frameworks. The United States enacted the Orphan Drug Act in 1983, promoting drug development through tax incentives, R&D support, and market exclusivity (*5*). The European Union implemented the Orphan Medicinal Products Directive in 2000, establishing cross-border collaboration mechanisms to harmonize drug approvals, clinical guidelines, and social security provisions (*6*). Japan and South Korea have also improved patient access to medications and diagnostic services through catalog recognition, research funding, and healthcare coverage (*7*). In contrast, China's policies began later but have gradually formed a system centered on catalogs, drugs, diagnostics, and coverage, in recent years (*8*). In 2018, China's National Health Commission released the "First Batch of Rare Disease Catalog", listing 121 diseases, which for the first time defined the scope of rare diseases and promoted drug development, standardized diagnostics, and medical insurance coverage (*9*).

In 2019, the Ministry of Finance and the General Administration of Customs introduced tax incentives to reduce drug prices (*10*). The same year saw the release of the "Guidelines for Diagnosis and Treatment of Rare Diseases (2019 Edition)", providing standardized clinical protocols for all 121 diseases and mandating their implementation by network hospitals (*9*). In 2023, the "Second Batch of Rare Disease Catalog" added 86 new conditions, bringing the total to 207, further expanding policy coverage and public awareness (*11*).

Existing research primarily focuses on policy content, international experiences, specific diseases, or regional cases, commonly employing literature reviews, comparative studies, and qualitative interviews (*12*). Systematic, standardized tools for quantitative assessment and visualization of national and provincial policies are rarely utilized. This study employs the Policy Modeling Consistency (PMC) index model and text mining methods to conduct quantitative analysis of policy texts at the national and regional levels in China. It aims to reveal the current status and shortcomings of China's rare disease policies, provide empirical evidence for policy optimization, and offer insights for the international community to understand China's rare disease governance practices.

2. Materials and Methods

2.1. Data sources

To ensure the representativeness and authority of the policy documents studied, this research employed the keyword "rare diseases" to conduct searches on the official websites of the State Council of the People's Republic of China and various ministries, as well as specialized policy databases such as "Peking University Law Database". Given the relatively limited number of specialized rare disease policies and China's overall late start in this field, the following criteria were applied for inclusion and exclusion:

Inclusion Criteria: *i*) Issuing authorities limited to the State Council of the People's Republic of China, ministries and commissions, provincial/municipal people's governments, and relevant government departments (*e.g.*, National Health Commission, local health commissions); *ii*) Document types restricted to plans, opinions, notices, and other policy-normative or directive documents; *iii*) Policy content must be closely related to the research theme. Exclusion criteria: *i*) Policy documents unrelated to the research theme; *ii*) Non-official documents such as bulletins, responses, or speeches.

China entered a phase of concentrated policy issuance in the rare disease sector starting in 2016, with a significant increase in policy density. This study selected policy documents issued between January 1, 2016, and June 1, 2025, as research texts. At the national level, a total of 19 policy documents were included (Table 1). At the local level, this study did not comprehensively cover all provinces but selected representative regions, such as those leading in rare disease diagnosis and treatment system development, medical insurance coverage, and drug accessibility, including a total of 13 policy documents (Table 2).

2.2. Policy text mining and analysis

Extracting the smallest analytical units highly relevant to the research theme from policy texts enhances the focus and scientific rigor of analysis, providing support for revealing core issues and evolutionary logic within rare disease policies. This study retained the smallest paragraphs highly relevant to the research theme from each policy text to construct a rare disease policy corpus. A combined qualitative and quantitative approach was employed to conduct a multi-perspective, multi-dimensional analysis of national and regional rare disease and drug-related policy texts. Following the compilation of national and regional policy texts, ROSTCM Content Mining 6.0 software was employed to perform text mining on 32 policies. This process involved filtering out non-specific policy indicators like "reduce" and "enhance", merging synonyms such as "evaluation" and "assessment", systematically removing function words and redundant general expressions, and generating a high-frequency word list (Table 3) and a semantic network diagram (Figure 1). This process identified policy focal points and intrinsic connections, establishing a data foundation for subsequent content analysis.

The high-frequency words reveal that rare disease policy texts predominantly focus on terms like "hospital", "patient", "treatment", "medical care", "clinical", and "disease". Policy emphasis centers on establishing diagnostic and treatment systems and providing patient healthcare services. Simultaneously, terms like "medicines", "drugs", "trials", and "review" reflect the state's high priority on drug R&D and regulatory approval processes. Conversely, terms related to financial burdens and social support, such as "guarantee" and "insurance", appear with lower frequency.

The semantic network diagram reveals that patients, clinical practice, and genetics occupy central positions within the social network. This indicates that clinical work, drug supply security, and genetic screening are the primary focuses of China's current rare disease policies. Furthermore, these three core high-frequency terms radiate out to multiple keywords, including treatment, disease, testing, mutation, drugs, and genetics. Consequently, enhancing the diagnostic and therapeutic standards for rare diseases, encouraging the market launch of rare disease drugs, and promoting innovative technological approaches represent the primary directions for China's current rare disease prevention, treatment, and policy safeguards.

Table 1. National-level policies on rare diseases in China

Policy Code	Policy Title	Issuing Authority	Date Issued
P1	Guiding Opinions on Promoting the Development of the Pharmaceutical Industry Chain (*37*)	General Office of the State Council	March 4, 2016
P2	Key Tasks for Deepening Healthcare System Reform in 2016 (*38*)	State Council	April 21, 2016
P3	Notice on Issuing the "Guidelines for Pharmaceutical Industry Development Planning" (*39*)	Ministry of Industry and Information Technology, National Development and Reform Commission, Ministry of Science and Technology	October 26, 2016
P4	Opinions on Deepening Reform of Review and Approval Systems to Encourage Innovation in Pharmaceuticals and Medical Devices (*40*)	State Council	October 8, 2017
P5	Notice on Publishing the First Batch of Rare Disease Catalog (*41*)	National Health Commission, Ministry of Science and Technology, Ministry of Industry and Information Technology	May 11, 2018
P6	Announcement on Optimizing Matters Related to Drug Registration Review and Approval (*42*)	National Medical Products Administration, National Health Commission	May 23, 2018
P7	Technical Guidance Principles for Accepting Overseas Clinical Trial Data for Drugs (*43*)	National Medical Products Administration	July 6, 2018
P8	Announcement on Issuing Guidance Principles for the Registration Review of Medical Devices Used in the Prevention and Treatment of Rare Diseases (*44*)	National Medical Products Administration	October 12, 2018
P9	Notice on Issuing the Diagnosis and Treatment Guidelines for Rare Diseases (2019 Edition) (*45*)	National Health Commission	February 27, 2019
P10	Notice on Establishing the National Rare Disease Diagnosis and Treatment Collaborative Network (*46*)	National Health Commission	February 12, 2019
P11	Notice on Value-Added Tax Policies for Rare Disease Medicines (*47*)	Ministry of Finance, General Administration of Customs, State Taxation Administration, National Medical Products Administration	February 20, 2019
P12	Notice on Implementing the Registration of Rare Disease Case Diagnosis and Treatment Information (*48*)	National Health Commission	October 10, 2019
P13	State Administration for Market Regulation (*49*)	State Administration for Market Regulation	March 30, 2020
P14	Technical Guidance Principles for Clinical Development of Rare Disease Drugs (*50*)	Drug Evaluation Center, National Medical Products Administration	December 31, 2021
P15	Guidance Principles for Natural History Studies in Rare Disease Drug Development (*51*)	Drug Evaluation Center, National Medical Products Administration	July 27, 2023
P16	Notice on Announcing the Second Batch of Rare Disease Catalog (*52*)	National Health Commission	September 18, 2023
P17	Technical Guidance Principles for Clinical Trials of Rare Disease Gene Therapy Products (*53*)	Drug Evaluation Center, National Medical Products Administration	January 12, 2024
P18	National Reimbursement Drug List for Basic Medical Insurance, Work Injury Insurance, and Maternity Insurance (2024 Edition) (*54*)	National Healthcare Security Administration, Ministry of Human Resources and Social Security	November 27, 2024
P19	Notice on Issuing Diagnosis and Treatment Guidelines for 86 Rare Diseases, Including Achondroplasia (2025 Edition) (*55*)	National Health Commission	June 17, 2025

Data Source: Compiled based on publicly available information from the official websites of the National Health Commission, the National Healthcare Security Administration, and other relevant authorities. For detailed references, see the bibliography at the end of this document.

Table 2. Policies related to rare diseases in typical regions

Region	Policy Code	Policy Title	Issuing Authority	Date Issued
Qingdao	P1	Qingdao Supplementary Medical Insurance Directory of Special Medicines, Special Medical Consumables, and Precision Diagnosis and Treatment Projects (*56*)	Qingdao Municipal Human Resources and Social Security Bureau	July 26, 2018
	P2	Notice of Qingdao Municipal Health Commission on Forwarding Document Lu Yibao Fa [2020] No. 84 Regarding Inclusion of Certain Rare Disease Specialty Medicines into Provincial Critical Illness Coverage (*57*)	Qingdao Municipal Medical Security Bureau, Qingdao Municipal Finance Bureau, Qingdao Municipal Health Commission	January 26, 2021
Shanghai	P3	Shanghai List of Major Rare Diseases (2025 Edition) (*58*)	Shanghai Municipal Health Commission and Four Other Departments	March 3, 2025
	P4	Shanghai Municipal Medical Security Regulations (*59*)	Standing Committee of the Shanghai Municipal People's Congress	December 31, 2024
Beijing	P5	Implementation Plan for Beijing's Pilot Zone Construction on Rare Disease Drug Coverage (Trial) (*60*)	Beijing Municipal Drug Administration and Five Other Departments	September 14, 2024
	P6	Capital Health Development Special Research Program on Rare Diseases (*61*)	Beijing Municipal Health Commission	May 27, 2025
Guangzhou	P7	Guangzhou Municipal Medical Security Development Plan for the 14th Five-Year Period (*62*)	Guangzhou Municipal Medical Security Bureau	December 28, 2021
Foshan	P8	Foshan Municipal Medical Assistance Measures (*63*)	Foshan Municipal Medical Security Bureau	February 28, 2025
	P9	Foshan City Catalog of Medicines, Therapeutic Foods, and Medical Institutions for Rare Disease Medical Assistance (2020 Edition) (*64*)	Foshan Municipal Medical Security Bureau	May 29, 2020
Jiangsu	P10	Jiangsu Province Implementation Plan for Rare Disease Diagnosis and Treatment (*65*)	Jiangsu Provincial Health Commission	April 2, 2019
	P11	Notice on Clarifying Matters Related to Diagnosis, Medication Treatment, and Benefit Eligibility for Rare Disease Medication Coverage Recipients (*66*)	Jiangsu Provincial Medical Security Bureau	July 6, 2021
Zhejiang	P12	Notice on Establishing the Zhejiang Province Rare Disease Medication Security Mechanism (*67*)	Zhejiang Provincial Medical Security Bureau, Zhejiang Provincial Department of Civil Affairs, Department of Finance	December 30, 2019
	P13	Notice on Strengthening Medical Security for Rare Diseases (*68*)	Zhejiang Provincial Department of Human Resources and Social Security, Zhejiang Provincial Department of Civil Affairs, Zhejiang Provincial Department of Finance, Zhejiang Provincial Health and Family Planning Commission	May 13, 2016

Data Source: Compiled based on publicly available information from the official websites of local health commissions, medical insurance bureaus, and other relevant authorities. For detailed references, please see the appendix at the end of this document.

2.3. PMC index model

The PMC index model was proposed by Estrad (*13*). Based on the Omnia Mobilis theoretical framework, it serves as an effective tool for measuring policy impact. This theory advocates prioritizing the influence of every relevant variable during policy evaluation. The PMC index model enables in-depth analysis of policy strengths and weaknesses across multiple dimensions, thereby facilitating quantitative assessment of policy texts.

2.3.1. Variable selection and configuration

Variable selection and indicator configuration are critical for policy analysis using the PMC Index Model. Based on high-frequency vocabulary and referencing existing

Table 3. High-frequency word list

No.	Term
1	Hospital
2	Patient
3	Treatment
4	Healthcare
5	Clinical
6	Disease
7	Gene
8	Drug
9	Diagnosis
10	Coverage
11	Mutations
12	Drugs
13	Neurological
14	Syndrome
15	Cell
16	Research
17	Institution
18	Rare
19	Abnormal
20	Test
21	Screening
22	Internal Medicine
23	Symptoms
24	Associated
25	Causes
26	Differential Diagnosis
27	Genetic
28	Metabolic
29	Management
30	Level
31	Pediatric
32	Disorder
33	Service
34	Developmental
35	Standard
36	Insurance
37	Protein
38	Immune
39	Endocrine
40	Diagnosis and Treatment
41	Data
42	System
43	Defect
44	Nation
45	Center
46	Development
47	Review
48	Severe
49	Quality
50	Chromosome

literature on the PMC Index Model construction, a quantitative evaluation system for China's rare disease policies was developed after expert consultation. This system comprises 9 primary variables and 32 secondary variables to comprehensively and specifically analyze China's rare disease policies (Table 4).

2.3.2. Calculation method for the PMC index model

Following Estrada's proposed calculation method for the PMC index model, the process primarily involves the following steps: First, assign values of 0 or 1 to secondary variables; second, calculate the scores for primary variables using Equation (3); finally, aggregate the scores of all primary variables for each policy using Equation (4).

$$X \sim N[0,1] \quad (1)$$

$$X = \{XR[0 \sim 1]\} \quad (2)$$

$$X_i = [\sum_{j=1}^{n} \frac{X_{ij}}{T(X_{ij})}] \quad (3)$$

$$PMC = \sum_{i=1}^{19} (X_i[\sum_{j=1}^{n} \frac{X_{ij}}{T(X_{ij})}] \quad (4)$$

Referencing existing evaluation metrics, policies are categorized into four grades: Perfect (7 ≤ PMC Index ≤ 9), Excellent (5 ≤ PMC Index < 6.99), Acceptable (3 ≤ PMC Index < 4.99), Poor (0 ≤ PMC Index < 2.99) (Table 5).

2.3.3. PMC surface plot construction

The PMC surface plot is constructed based on a 3 × 3 PMC matrix, with the specific construction method shown in Equation (5). The PMC surface plot visually presents policy evaluation results more intuitively. It substitutes the mean values of each policy's primary variables, imports the data into Excel, and generates the overall mean PMC surface plot for the policy. In this surface plot, 1, 2, and 3 represent the horizontal coordinates of the matrix, while Series 1, 2, and 3 denote the vertical coordinates. The PMC surface plot uses different colors to distinguish index scores, enabling visual assessment of policy refinement. Convex regions indicate higher scores for corresponding evaluation indicators, while concave regions reflect lower scores.

$$PMC = \begin{Bmatrix} x1 & x2 & x3 \\ x4 & x5 & x6 \\ x7 & x8 & x9 \end{Bmatrix} \quad (5)$$

3. Results

3.1. Overall evaluation of national policies

By constructing an input-output analysis model for national rare disease policies and calculating the PMC Index, the rationality and maturity of the policy system can be systematically assessed, providing empirical evidence for optimizing policy design and enhancing implementation effectiveness. Based on the variable system of national rare disease policies, an input-output analysis model was developed to calculate the PMC Index and evaluation grades for 19 policies (Table 6). The average PMC Index across all policies was 5.31, earning an Excellent rating. This outcome reflects China's significant progress in rare disease policy development, demonstrating overall rational policy design with a stratified rating distribution: 2 policies rated Perfect, 8

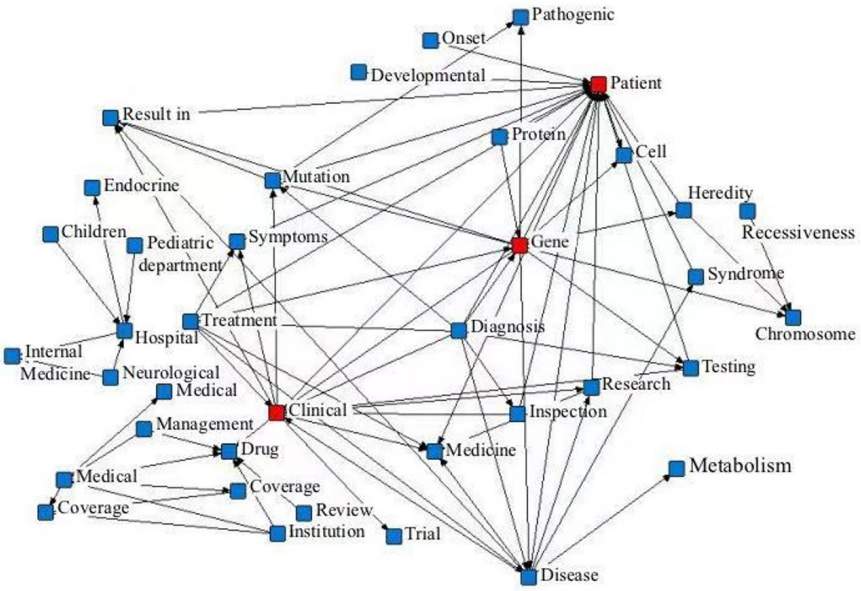

Figure 1. Semantic network diagram.

rated Excellent, and 9 rated Acceptable, with no Poor ratings. China's rare disease policies have thus reached a mature stage.

A radar chart was constructed using the mean values of the 19 primary variables for rare disease policies (Figure 2). Examining specific policy dimensions reveals higher average scores for Policy Nature (X2) and Policy Outcome Evaluation (X4). Currently, China's rare disease policies demonstrate strong strategic planning and outcome orientation, with clear long-term objectives. The strategic planning dimension (X2,1) indicates that policies generally provide a guiding framework for future development. Additionally, the high score for Policy Functionality (X8) suggests that China's policies emphasize oversight and accountability during implementation, effectively driving policy execution. Issuing Authority (X1) reflects that China's rare disease policies are primarily developed through multi-departmental leadership, with active participation from various government agencies.

The incentive measures (X5) scored the lowest, indicating that China's policy measures in areas such as R&D funding support and clinical trial incentives are relatively inadequate, affecting the development and market access of innovative drugs. Furthermore, the scores for patient support (X6) and social support (X9) were 0.42 and 0.39, respectively, reflecting that patient protection and social integration mechanisms remain weak. In particular, social integration measures have not effectively eliminated the social discrimination and employment barriers faced by rare disease patients.

3.2. Analysis of typical national policy cases

By comparing high-scoring and low-scoring policies, we can reveal differences in strategic levels, coverage scope, and implementation effectiveness in policy design. This provides a reference for optimizing the rare disease policy system and formulating more targeted and scientifically grounded policies. Among policies issued at the national level, we selected the two highest-scoring and the two lowest-scoring policies for case analysis, constructing a surface plot accordingly (Figure 3, and Supplementary Figure S1, *https://www.irdrjournal.com/action/getSupplementalData.php?ID=283*). Policy P1, "Guiding Opinions on Promoting the Development of the Pharmaceutical Industry Chain", focuses on strengthening technological innovation, advancing industrial restructuring and cluster development, enhancing production and supply capabilities for generic drugs, essential medicines, and clinically urgent drugs; improving quality standards and testing systems; establishing fast-track review and market access channels for shortage drugs and innovative drugs; and aligning the registration system with international standards. Overall, it exhibits a high strategic level, broad coverage, and a comprehensive system. Policy P4, "Opinions on Deepening the Reform of the Review and Approval System to Encourage Innovation in Pharmaceuticals and Medical Devices", explicitly proposes deepening the review and approval system and establishes a regulatory framework tailored to innovative drugs. It enables accelerated review for urgently needed medicines and simultaneous submission of new drugs from overseas, significantly shortening time-to-market. Its incentive measures are relatively comprehensive in scope.

However, Policy P11, "Notice on Value-Added Tax Policies for Rare Disease Drugs", focuses solely on tax reductions and serves as a supplementary, supporting measure. Policy P9, "Notice on Issuing the Diagnosis

Table 4. National-level policy evaluation system

Primary Variable	Code	Secondary Variable Name	Secondary Variable Evaluation Criteria	Indicator Source
Issuing Agency (X_1)	X_{11}	State Council	Whether the policy issuing body is the State Council itself.	Modified from Jin Chen's (*69*) article
	X_{12}	General Office of the State Council	Whether the policy issuing body is the General Office of the State Council.	
	X_{13}	State Council Constituent Departments (Ministries and Commissions)	Whether the policy issuing body is a ministry-level agency.	
	X_{14}	State Council Directly Affiliated Agencies	Whether the policy issuing body is a directly affiliated agency.	
	X_{15}	Internal Departments and Subordinate Units	Whether the policy issuing body is a subordinate unit.	
	X_{16}	Multi-Agency Joint Release	Whether the policy is jointly issued by two or more of the above agencies.	
Policy Nature (X_2)	X_{21}	Strategic Planning	Whether the policy proposes long-term goals, development directions, or top-level design frameworks.	Modified from Ruiz Estrada (*13*) and Zhang Yong'an (*70*) articles
	X_{22}	Guidance and Coordination	Whether the policy provides action guidelines, cross-department collaboration mechanisms, or multi-party coordination requirements.	
	X_{23}	Supervision and Control	Whether the policy specifies regulatory bodies, accountability mechanisms, or mandatory constraint clauses.	
	X_{24}	Prediction and Evaluation	Whether the policy includes risk warnings, effect evaluations, or dynamic adjustment mechanisms.	
Drug Security (X_3)	X_{31}	Market Access and Incentives	Focuses on how to encourage pharmaceutical companies to research, produce, and quickly bring drugs to market.	Modified from rare disease policy texts
	X_{32}	Medical Insurance Payment and Accessibility	Focuses on how to ensure patients have access to and can afford these drugs.	
Policy Outcome Evaluation (X_4)	X_{41}	Clarity of Goal Setting	Whether the goals set by the policy are clear.	Modified from Wang Jinfu (*71*) and Zhang Li (*72*) articles
	X_{42}	Rationality and Sufficiency of Basis	Whether the basis for policy formulation is sufficient.	
	X_{43}	Practicality of Planning	Whether the policy planning is feasible to implement.	
Incentive Measures (X_5)	X_{51}	Resource Sharing	Whether the policy relates to resource sharing.	Modified from Zhang Yong'an's (*70*) article
	X_{52}	Institutional Guarantees	Whether the policy relates to institutional guarantees.	
	X_{53}	Talent Development	Whether the policy relates to talent development.	
	X_{54}	Finance and Taxation	Whether the policy relates to finance and taxation.	
	X_{55}	Clinical Trial Support	Whether the policy supports the conduct of clinical trials.	
	X_{56}	R&D Funding	Whether the policy relates to research and development funding support.	
	X_{57}	Other	Whether the policy relates to other incentive measures.	
Patient Support (X_6)	X_{61}	Diagnosis, Treatment, and Survival Support	Whether it provides support directly benefiting individual patients, such as diagnosis, medication, rehabilitation, and living subsidies.	Modified from rare disease policy texts
	X_{62}	Rehabilitation and Social Adaptation Support	Whether it provides psychological counseling, vocational skills training, social adaptation guidance, *etc*., for individual patients to enhance their social participation ability.	

Table 4. National-level policy evaluation system (continued)

Primary Variable	Code	Secondary Variable Name	Secondary Variable Evaluation Criteria	Indicator Source
Technological Innovation (X_7)	X_{71}	R&D Drive and Translation	Measures whether the policy systematically incentivizes the translation of cutting-edge basic research into clinical applications.	Modified from rare disease policy texts
	X_{72}	Early Screening System Construction	Measures the policy's efforts in building a comprehensive, life-cycle rare disease prevention and early detection network.	
	X_{73}	Accelerated and Inclusive Review	Measures the innovation and flexibility of the drug regulatory system.	
	X_{74}	Data Empowerment and Collaboration	Measures the policy's support for scientific research infrastructure and collaborative ecosystems.	
Policy Function (X_8)	X_{81}	Supervision and Constraints	Whether it supervises and constrains rare disease treatment.	Based on Xuan Tianhui's (*73*) article, modified
	X_{82}	Standardization and Guidance	Whether it standardizes and guides rare disease treatment.	
	X_{83}	Quality Improvement	Whether it improves the quality of rare disease treatment.	
Social Support (X_9)	X_{91}	Information Platform and Data Construction	Whether a unified rare disease information sharing system has been established.	Modified from rare disease policy texts
	X_{92}	Emergency Assistance and Medical Security	Whether a rare disease acute episode treatment channel has been established.	
	X_{93}	Rights Protection and Anti-Discrimination	Whether it explicitly prohibits genetic discrimination in employment/education/insurance fields.	
	X_{94}	Other	Whether it includes other macro-level social support policies not covered above.	

Table 5. Rare disease policy evaluation grades

Policy modeling consistency (PMC) index	0–2.99	3.0–4.99	5.0– 6. 99	7.0–9.0
Evaluation Grades	Poor	Acceptable	Excellent	Perfect

Table 6. Policy modeling consistency (PMC) index of national-level rare disease policy documents

Policy Code	X1	X2	X3	X4	X5	X6	X7	X8	X9	PMC Index	Ranking	Policy Level
P1	0.83	1.00	1.00	1.00	1.00	0.50	0.75	1.00	0.75	7.83	1	Perfect
P2	1.00	0.75	0.50	0.67	0.14	0.00	0.25	1.00	0.25	4.56	14	Acceptable
P3	1.00	1.00	0.50	1.00	0.43	0.00	0.75	1.00	0.50	6.18	5	Excellent
P4	1.00	1.00	1.00	1.00	0.57	0.50	0.75	1.00	0.25	7.07	2	Perfect
P5	1.00	0.50	1.00	1.00	0.14	0.50	0.25	1.00	0.50	5.89	6	Excellent
P6	0.92	1.00	0.50	1.00	0.71	0.50	0.50	1.00	0.75	6.88	3	Excellent
P7	0.50	0.25	0.50	1.00	0.29	0.00	0.75	0.67	0.25	4.20	17	Acceptable
P8	0.50	0.25	0.50	1.00	0.14	0.50	0.50	1.00	0.00	4.39	16	Acceptable
P9	0.67	0.50	1.00	1.00	0.14	0.50	0.00	1.00	0.25	5.06	9	Excellent
P10	0.67	0.75	0.50	1.00	0.57	0.50	0.50	1.00	0.75	6.24	4	Excellent
P11	0.87	0.25	0.00	1.00	0.29	0.50	0.00	1.00	0.25	4.16	18	Acceptable
P12	0.67	0.25	0.50	1.00	0.29	0.50	0.00	1.00	0.25	4.46	15	Acceptable
P13	0.50	0.50	0.00	1.00	0.29	1.00	0.50	1.00	0.25	5.04	10	Excellent
P14	0.33	0.75	0.00	1.00	0.86	0.50	0.50	1.00	0.50	5.44	8	Acceptable
P15	0.33	0.50	1.00	1.00	0.29	0.50	0.25	0.67	0.50	5.03	11	Excellent
P16	0.67	0.25	1.00	1.00	0.00	0.50	0.25	0.67	0.25	4.58	13	Acceptable
P17	0.33	0.50	0.50	0.67	0.14	0.50	0.75	1.00	0.25	4.64	12	Acceptable
P18	1.00	0.50	1.00	1.00	0.29	0.50	0.25	0.67	0.50	5.70	7	Excellent
P19	0.67	0.50	0.00	1.00	0.50	0.00	0.00	0.33	0.50	3.50	19	Acceptable
Mean	0.71	0.58	0.58	0.96	0.37	0.42	0.39	0.89	0.39	5.31	—	Excellent

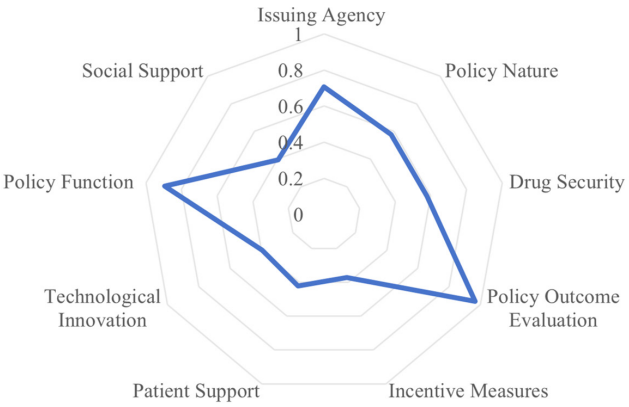

Figure 2. National rare disease policy modeling consistency (PMC) mean radar chart.

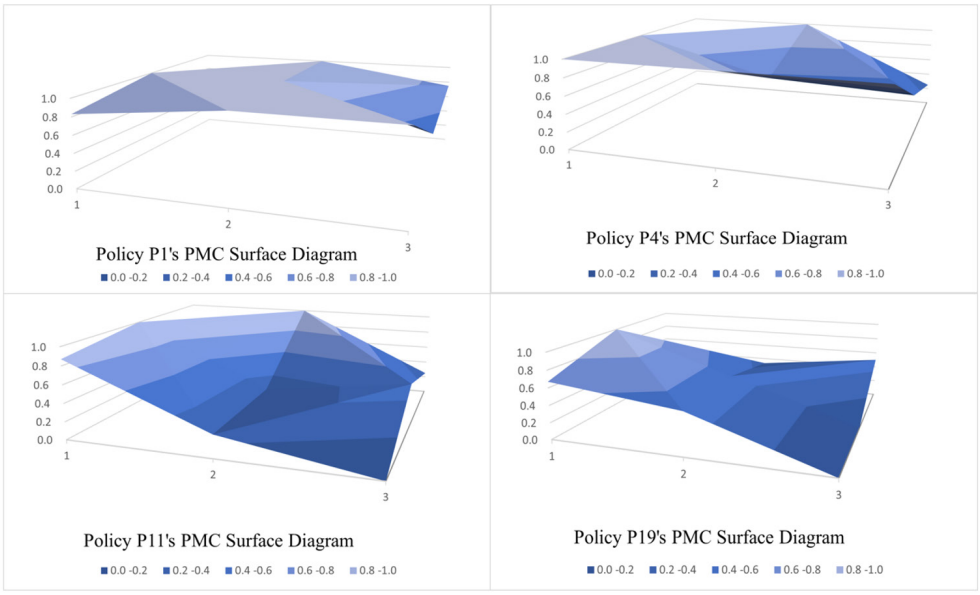

Figure 3. Policy modeling consistency (PMC) surface plot of national rare disease policies.

and Treatment Guidelines for 86 Rare Diseases Including Achondroplasia (2025 Edition)", primarily addresses medical practice and has limited coverage, resulting in a lower score.

3.3. Overall evaluation of regional policies

The analysis of provincial policy cases reveals the strengths and weaknesses of China's provincial-level regional policies by assessing the design characteristics and implementation effectiveness of rare disease policies across regions. This provides reference for optimizing local policy systems, promoting differentiated policy innovation, and advancing scientific decision-making. Based on the variable system of provincial rare disease policies, an input-output analysis model was developed to calculate the PMC index and evaluation grade for 13 policies (Table 7). The average PMC index for all policies was 4.94, with policy ratings showing a stratified distribution: 4 policies rated Excellent, 9 rated Acceptable, and none rated Poor. This indicates that the sampled policies were generally well-designed, with rare disease policies in typical regions becoming increasingly refined. This reflects an overall pattern where, under national strategic guidance, localities are exploring distinctive models.

A radar chart was constructed using the mean values of the primary variables for the 13 rare disease policies (Figure 4). Analysis of specific dimensions reveals that policy nature (X2) generally scored highly, particularly in Beijing and Shanghai. Their policies demonstrate strong strategic and long-term planning capabilities, reflecting robust top-level design and organizational coordination by governments in advancing rare disease policies. Additionally, high scores for policy functionality (X8) indicate these regions possess strong oversight mechanisms and implementation capacity, ensuring policy execution and enforcement.

Incentives (X5) and Patient Support (X6) represent weaknesses in most regional policies. The average score

Table 7. Policy modeling consistency (PMC) index of rare disease policy documents in representative regions

Policy Code	X1	X2	X3	X4	X5	X6	X7	X8	X9	PMC Index	Ranking	Policy Level
P1	0.60	0.25	1.00	1.00	0.27	0.00	0.00	1.00	0.25	4.37	9	Acceptable
P2	0.80	1.00	1.00	1.00	0.14	0.50	0.25	1.00	0.25	5.94	3	Excellent
P3	1.00	0.25	0.00	0.67	0.00	0.50	0.50	1.00	0.25	4.17	12	Acceptable
P4	1.00	1.00	1.00	1.00	0.29	0.50	0.25	1.00	0.25	6.29	2	Excellent
P5	1.00	1.00	1.00	0.67	0.43	0.50	0.50	1.00	0.25	6.35	1	Excellent
P6	1.00	0.25	0.50	1.00	0.29	0.00	0.67	1.00	0.25	4.96	6	Acceptable
P7	0.80	0.50	0.00	1.00	0.43	0.00	0.25	1.00	0.25	4.23	11	Acceptable
P8	0.60	0.25	0.50	0.67	0.29	0.50	0.00	1.00	0.50	4.31	10	Acceptable
P9	0.60	0.25	0.50	1.00	0.00	1.00	0.00	0.33	0.25	3.93	13	Acceptable
P10	0.60	1.00	0.00	1.00	0.57	0.50	0.25	1.00	0.25	5.17	5	Excellent
P11	0.60	0.75	1.00	0.67	0.14	0.50	0.00	0.67	0.25	4.58	8	Acceptable
P12	0.80	1.00	1.00	0.67	0.14	0.50	0.00	1.00	0.25	5.36	4	Acceptable
P13	0.80	0.25	1.00	0.67	0.14	0.50	0.00	1.00	0.25	4.61	7	Acceptable
Mean	0.78	0.60	0.65	0.85	0.24	0.42	0.21	0.92	0.27	4.94	-	Acceptable

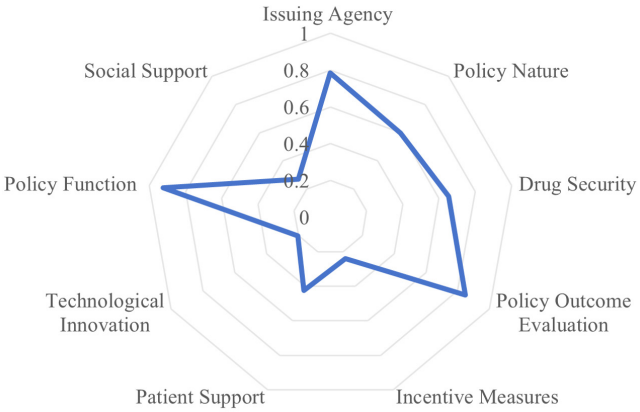

Figure 4. Radar chart of average policy modeling consistency (PMC) for rare disease policies in representative regions.

for local policy X5 (Incentives) is 0.24, significantly lower than the national policy average of 0.37. Both X6 (Patient Support) scores are low, though local measures demonstrate greater operational feasibility—such as Qingdao's specialized drug coverage program.

Shanghai, Beijing, and Zhejiang exhibit more systematic policy design. These economically developed regions with concentrated medical resources generally achieve higher policy scores. Jiangsu and Qingdao perform well in medical insurance payments and treatment protocols — Jiangsu's implementation plan specifies diagnostic pathways. At the same time, Qingdao explores incorporating certain specialty drugs into major illness insurance, though research and incentive measures remain inadequate. Foshan and Guangzhou focus their policies on medical assistance and insurance planning, emphasizing a "safety net" function, but lack systematic design and have room for refinement.

3.4. Analysis of typical regional policies

Since 2005, Shanghai, Qingdao, and Zhejiang Province have successively explored rare disease drug coverage models independent of the basic medical insurance system, which have, to some extent, alleviated patients' payment burden (*14*). Shanghai stands as one of China's pioneers in addressing the unique needs of the rare disease community, consistently leading domestic efforts in rare disease healthcare coverage. Both Shanghai and Beijing score highly in innovative drug support and patient protection policies. Shanghai's release of the Shanghai Catalogue of Major Rare Diseases (2025 Edition) and the Shanghai Medical Security Regulations not only enhances drug accessibility but also clarifies social support and policy safeguards for patients. Beijing, through its Capital Health Development Rare Disease Research Special Project policy, provides robust financial backing for rare disease research and drug development, ensuring policy effectiveness. Qingdao has established a multi-tiered healthcare security system prioritizing rare disease patients, incorporating over 20 rare diseases into outpatient critical illness management, and implementing specialized coverage for three rare disease-specific drugs (*15*). Guangzhou has constructed a multi-tiered support system covering diagnosis, treatment, medical insurance, and assistance. It features a Major Complex and Rare Disease Diagnosis and Treatment Center integrating Chinese and Western medicine, while leveraging provincial collaborative network hospitals like Guangzhou Medical University Women and Children's

Medical Center to promote early screening and diagnosis. Beyond implementing the national medical insurance catalog, it supplements coverage with insurance that does not restrict pre-existing conditions. Foshan City has included all diseases listed in the "First Batch of Rare Diseases Catalog" within its medical assistance coverage, extending this support to cover outpatient specialty and therapeutic food expenses for rare disease patients beyond basic medical insurance. The proactive explorations in rare disease medical coverage in regions like Zhejiang and Jiangsu provide practical models for achieving nationwide coverage, offering highly valuable reference points (Supplementary Table S1 and Supplementary Figure S2, *https://www.irdrjournal.com/action/getSupplementalData.php?ID=283*).

4. Discussion and recommendations

4.1. Comparison and implications of national and local rare disease policies

Based on the quantitative results of the Policy Management Center (PMC) analysis, differences exist between national and local policies in terms of functional positioning and implementation pathways. These variations not only reveal the structural characteristics of China's rare disease policy framework but also provide entry points for subsequent institutional optimization.

Among the 32 policy documents evaluated, 19 were national-level policies and 13 were local-level policies. Overall, national policies exhibit broad coverage and high institutionalization, primarily focusing on foundational institutional designs such as establishing drug directories, drug registration and review, medical insurance reimbursement, and diagnostic and treatment guidelines. They perform notably well in the Institutional Foundation (X2) and Policy Tool Completeness (X3) dimensions of the PMC index. Local policies, however, are more targeted and exploratory, often focusing on supplementary medical insurance payments, establishing relief funds, expanding patient assistance channels, and innovating multi-tiered security systems. They demonstrate certain advantages in the areas of safeguard measures (X4) and incentive mechanisms (X5).

This divergence reflects the complementary relationship between national and local policies in their functional roles. National-level policies are responsible for top-level design and establishing institutional frameworks, providing unified standards and fundamental safeguards for rare disease governance. While local policies conduct differentiated explorations based on economic conditions and social resources, they offer more targeted support to patients. However, national policies still need to enhance their specificity in implementation, while local policies, constrained by fiscal capacity and institutional authority, exhibit weaker coverage and sustainability (*16*). Future policy optimization should establish tighter coordination mechanisms between national and local levels. The national government should strengthen institutional safeguards and resource coordination. In contrast, local governments should serve as "policy testing grounds", accumulating practical experience and feeding it back to the national level to drive the continuous improvement and balanced development of the rare disease policy system.

4.2. Pathways for improving institutional frameworks and policy safeguards

Since the release of the "Guidelines for Diagnosis and Treatment of Rare Diseases (2019 Edition)", China became the first country to define rare diseases through a catalog system, laying a crucial institutional foundation for subsequent policy development and drug review and approval processes. However, comparisons with international experiences reveal gaps in China's policy safeguarding system, particularly in policy stability, interdepartmental coordination, and legal enforceability. Current policies primarily rely on departmental regulations and technical guidelines, lacking unified national legislation. This leads to fragmented implementation and inconsistent standards across regions. Additionally, oversight and dynamic evaluation mechanisms remain underdeveloped, with some localities exhibiting weak enforcement and inadequate interdepartmental coordination during implementation (*17*).

To address these issues, it is recommended to enhance the rare disease policy framework through legislative and institutional measures (*18*). At the macro level, national efforts should advance the enactment of a "Rare Diseases Law" or other specialized legislation (*19*), incorporating fundamental patient healthcare rights, orphan drug R&D incentive mechanisms, rules for establishing and allocating dedicated rare disease funds, interdepartmental collaboration protocols, and information-sharing systems into the legal framework. At the local level, tailored approaches should be adopted to establish robust policy implementation and oversight mechanisms, considering China's context of substantial economic scale but relatively low per capita income (*20*). Transparency and fairness can be ensured by involving third-party institutions and social organizations. Throughout this process, policies should be optimized and revised based on implementation outcomes. This institutional design enhances policy transparency and accountability while providing evidence-based support for dynamic adjustments, thereby fostering a virtuous cycle of "legislation-implementation-evaluation-revision".

4.3. Regional disparities and optimization of policy coordination mechanisms

Regional disparities have become a prominent issue in the practical implementation of China's rare disease policies. Economically developed regions have demonstrated a leading advantage in institutional development, research investment, and innovation safeguards (*21*). However, policy frameworks in small and medium-sized cities and underdeveloped areas remain weak. Local initiatives heavily depend on regional economic development levels, policy implementation speed, and local sociocultural traditions, preventing them from extending assistance beyond the nationally mandated drug directory (*22*). Within the framework of basic medical security, benefit levels under basic medical insurance, critical illness insurance, and medical assistance remain constrained by existing policy structures, making it difficult to implement special protective policy adjustments for rare disease patients (*17*). This results in significant regional disparities in diagnostic accessibility, insurance reimbursement rates, and assistance levels for rare disease patients. Concurrently, China's household registration system and local coordination mechanisms further exacerbate coverage inequities, creating substantial gaps in access to rare disease medical services and pharmaceutical support across different regions.

To enhance policy universality and sustainability, the national level should promote cross-regional policy coordination and resource integration, providing preferential support to underdeveloped areas to improve local diagnostic capabilities and coverage levels. Establishing a unified national rare disease collaboration network would enable cross-regional sharing of research platforms, clinical resources, and referral networks, thereby institutionally facilitating the decentralization of superior resources (*23*). Thus strengthening fiscal burden-sharing and technical support between central and local governments, while guiding local authorities to develop tailored, localized measures. Promoting a "best practices" model that integrates Shanghai's institutional innovations, Beijing's research strengths, and multi-tiered coverage experiences from Guangdong, Qingdao, and other regions—while institutionalizing and scaling these approaches within a national framework—can progressively narrow regional disparities. This shift from "isolated experimentation" to "networked collaboration" in rare disease governance enhances overall fairness and sustainability, offering valuable pathways for refining China's distinctive health governance model.

4.4. Incentive system development and strategies to promote orphan drug R&D

Rare disease drug development is central to improving patient care and drug accessibility. Despite recent national policies encouraging innovation, support remains insufficient in R&D funding, clinical trial incentives, and market access for innovative drugs. This results in weak corporate R&D motivation, clinical trial bottlenecks, and inefficient drug commercialization. High drug prices reduce patient access, while low prices may deter innovation, ultimately compromising patient outcomes (*24*). Systematic legal and policy incentives are crucial drivers for orphan drug development. The U.S. Orphan Drug Act significantly boosted new drug development output through market exclusivity, tax incentives, and R&D funding. The EU similarly advanced orphan drug innovation *via* multinational collaboration mechanisms, centralized approval processes, and financial incentives. These experiences demonstrate that combining institutional incentives with market mechanisms can stimulate corporate innovation in rare disease areas.

China should establish a more comprehensive national R&D system and build a national rare disease research and clinical data sharing platform. This platform should enable standardized sharing of patient registries, real-world evidence, and multicenter clinical trial data, thereby reducing research costs and enhancing R&D efficiency (*18*). Establishing priority review and approval pathways, coupled with granting orphan drugs a certain period of market protection, would improve the feasibility and economic returns of drug development and market launch. A dedicated talent development program for rare diseases should be launched, offering research funding preferences and incentives to young scientists and clinicians to encourage greater participation in this field. National-level policy design should refine supportive measures, enhance corporate social responsibility, and ensure the production and supply accessibility of rare disease medications (*25*). International collaboration should be promoted by sharing technologies, evaluation standards, and clinical research evidence with global orphan drug agencies to address data gaps and inconsistencies in evaluation during R&D. Concurrently, China should enhance public awareness of rare diseases, stimulate societal engagement, and incentivize pharmaceutical companies to engage in orphan drug development (*26*). These measures will progressively establish a comprehensive system encompassing financial support, institutional incentives, and talent cultivation. This framework will foster a conducive regulatory environment for orphan drug research and development, thereby enhancing innovation capacity and clinical accessibility for rare disease medications (*27*).

4.5. Establishing patient support systems and specialized insurance for rare diseases

Rare diseases pose significant challenges not only in medical and physiological aspects but also profoundly impact patients' and their families' economic and social lives. Although some rare diseases have been included in medical insurance coverage, the scope and level of protection remain limited. The prolonged treatment

cycles and need for multiple medications result in persistent financial burdens, placing heavy economic and psychological strain on patients during long-term care (*28*). A study assessing the accessibility of definitive diagnosis for rare diseases among Chinese adults revealed that approximately 72.97% of patients received misdiagnoses. On average, patients endured 4.30 misdiagnoses and visited 2.97 hospitals before receiving a correct diagnosis (*27*). In China, very few rare disease patients possess commercial insurance, with the majority lacking any commercial coverage (*27*). Few commercial insurers are willing to include high-cost, low-volume, rare disease-specific medications within their coverage scope (*29*).

In addressing this issue, the United States, the European Union, Japan, and Taiwan have implemented various financing models, including commercial insurance, social charitable funding, medical assistance programs, and government subsidy funds (*30*). Therefore, to further enhance the patient support system, it is essential to expand medical insurance coverage and increase reimbursement rates for rare disease medications to meet patients' survival and developmental needs better. Incorporating rare diseases into basic medical insurance and critical illness insurance can alleviate patients' financial burdens. A dedicated rare disease protection fund should be established under critical illness insurance, achieving comprehensive provincial coordination for this specific category. Drawing from Zhejiang's experience, annual contributions to this fund could be allocated from the critical illness insurance pool within each coordinated region. However, given the limited capacity of government medical insurance funds, supplementary security mechanisms like commercial insurance are crucial for addressing the high costs associated with rare disease treatment (*31*). Special insurance plans should be established to cover treatment and long-term management expenses for rare diseases, and promote the establishment of specialized psychological support and rehabilitation service centers to provide patients with counseling and social adaptation guidance. Social organizations should be encouraged and public welfare foundations be engaged in patient services, forming a multi-tiered support network. Expanding medical insurance coverage, establishing a dedicated rare disease fund, gradually increasing reimbursement rates, and actively providing patients with living allowances, rehabilitation subsidies, and long-term medication assistance to alleviate family burdens is necessary. Through concerted efforts from multiple stakeholders, enhancing patient support capabilities is essential(*32*).

4.6. Synergistic effects of social support systems and multi-stakeholder collaboration

Rare disease patients face not only medical challenges but also heightened survival pressures due to inadequate social support. While the government has provided basic medical coverage to some extent, social-level support remains insufficient, particularly in areas like social adaptation and employment, where patients encounter significant difficulties (*33*). China's current social support system remains in its early stages, with low social participation rates among rare disease patients. Constrained by societal misconceptions, educational barriers, and employment discrimination, patients endure significant psychological and financial pressures (*34*). For instance, some EU countries utilize charitable funds and social security programs to provide living allowances and rehabilitation support for rare disease patients, while the United States encourages collaborations between social organizations and corporations to advance rare disease research and drug development, promoting broader drug accessibility.

Therefore, a unified national rare disease information platform should be established to facilitate the sharing of patient registries, diagnostic guidelines, drug directories, and other critical information. Enhanced big data applications should support precision in scientific research and policy formulation. Concurrently, an emergency "green channel" should be created for acute episodes of rare diseases, ensuring priority treatment during emergency care and hospitalization. A temporary relief fund should be established to provide safety-net coverage for sudden high medical expenses. Legislation should explicitly prohibit discrimination in education, employment, and insurance. Public awareness campaigns and science education should be promoted to enhance public understanding of rare diseases and reduce stigma. Charitable organizations, public welfare funds, and social enterprises should be encouraged to participate in rare disease patient assistance, forming a multi-faceted social support system (*35*) that pools diverse resources to address medication accessibility for rare disease patients collectively.

4.7. Limitations of the study

This study analyzed the shortcomings of existing policies. However, upon examining the content, it was found that the research exhibits the following limitations in terms of sample coverage, methodological constraints, and practical evaluation. At the macro level, the representativeness of regional samples is limited. The study selected only 13 policy documents at the local level, concentrated in typical regions such as Shanghai, Beijing, and Qingdao. Compared to China's 34 provincial-level administrative regions, the coverage of these 13 local documents is narrow, making it difficult to fully reflect the diversity of local policies. The selected regions predominantly represent economically developed cities or provinces with concentrated healthcare resources. This selection bias may lead to

overly optimistic evaluation outcomes, failing to fully reflect the genuine gaps in rare disease policies within central, western regions, or areas with weaker healthcare infrastructure.

Second, while the PMC index model enables quantitative evaluation, its methodology inherently involves certain simplifications. The selection of secondary variables relies on expert consultation, and the assignment of variable values (0 or 1) carries a degree of subjectivity. Any discrepancies in interpreting and understanding policy provisions during scoring will directly impact the final score. Furthermore, when calculating primary variable scores, the model typically assumes equal importance across all secondary variables without employing weighted scoring. In practice, however, a fiscal subsidy policy may carry significantly greater weight and provide stronger policy support than a mere work notice. Such weighting differences cannot be captured by simple binary scoring.

Finally, the disconnect between policy texts and implementation outcomes is significant. Research primarily focuses on the consistency and structural completeness of policy texts rather than their actual effectiveness. The PMC model evaluates the textual quality of policies themselves. However, certain high-scoring policies (such as those rated P1 or P4 as perfect) may face practical challenges in implementation, including difficulties in cross-departmental coordination and inadequate funding allocation. Empirical analysis of such real-world outcomes remains limited in this paper.

5. Conclusion

This paper employs the Policy Modeling Consistency (PMC) index model and text mining methods to conduct a systematic quantitative evaluation of rare disease-related policies issued at the national level (19 policies) and in representative regions (13 policies) in China from 2016 to 2025. Regarding research methods and tools, ROSTCM 6.0 software was used to extract high-frequency vocabulary, identifying policy focal points primarily concentrated on "hospitals", "patients", "treatment", "clinical", and "medicines". An evaluation framework was constructed comprising nine primary variables—issuing authority, policy nature, drug coverage, outcome evaluation, incentive measures, patient support, technological innovation, policy function, and social support—along with 32 secondary variables. Policy strengths, weaknesses, and refinement levels were visually represented through PMC surface plots and radar charts.

Findings revealed high scores for policy nature (X2), policy function (X8), and outcome orientation (X4), indicating robust strategic planning and regulatory rigor. Conversely, incentive measures (X5), patient support (X6), and social support (X9) emerged as weak points with generally low scores, revealing deficiencies in R&D funding, social integration, and long-term security mechanisms. Regionally, developed areas like Shanghai, Beijing, and Zhejiang exhibited more systematic policy design, while Guangzhou and Foshan emphasized the safety net function of medical insurance.

Finally, the article recommends establishing a comprehensive policy framework covering R&D, diagnosis/treatment, medical insurance, and social support. This includes strengthening coordinated legislation to advance the Rare Disease Law, incorporating patient rights, orphan drug R&D incentives, and dedicated fund establishment into the legal system. Enhancing incentives and support requires building a national R&D system and information-sharing platform, while implementing market protection periods and fiscal/tax incentives for orphan drugs. Furthermore, regional coordination should be optimized to narrow coverage disparities, promote "best practice" models, and enable cross-regional sharing of research resources and clinical data. A multi-tiered insurance system should be developed, including a dedicated rare disease fund, while encouraging participation from commercial insurers and social welfare organizations to alleviate patients' financial burdens.

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