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Review

Assessment of Orphan Drug Accessibility in Kazakhstan: Comparative Analysis of Different Public Healthcare System Models

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Abstract

Orphan diseases remain a significant challenge for healthcare systems due to the high cost of treatment. The objective of this review is to compare international models of orphan drug accessibility and identify approaches that may be applicable to Kazakhstan.

A literature search was conducted for publications from 2015-2025 in PubMed, Scopus, Web of Science databases, and official WHO and EU documents. Studies with data on drug accessibility, financing mechanisms, and regulations were included. The selection process followed PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) guidelines.

Germany employs simplified Health Technology Assessment (HTA) procedures and automatic recognition of orphan drug benefits. France applies fixed tariffs and expedited reimbursement. In the USA, the Orphan Drug Act stimulates innovation but challenges of equal access persist. Japan and South Korea have implemented universal insurance and risk-sharing agreements. Russia operates centralized procurement and the Circle of Good fund. In Kazakhstan, 205 drugs are included in formularies, but barriers remain: limited neonatal screening, registration delays, and regional disparities.

International experience indicates the potential of implementing specialized HTA procedures, expanding screening programs, and developing expert centers. This will improve orphan disease treatment accessibility and maintain healthcare system sustainability.

Keywords: orphan diseases, drug accessibility, HTA, Kazakhstan.

1. Introduction

The problem of providing pharmaceutical access to patients with orphan diseases has become particularly acute in recent decades. The 2024 international definition establishes rarity criteria as ≤1 case per 2,000 individuals [1], yet behind these figures stand millions of patients — from 262.9 to 446.2 million people globally, with the majority of cases (80%) having a genetic etiology [2].

The paradox of orphan drugs lies in the fact that small patient populations face disproportionately high barriers to therapy access. Pharmaceutical companies must recoup enormous development costs from an extremely limited market, resulting in astronomical prices for finished products [3]. Different countries seek

their own solutions to this dilemma—from market incentives in the USA to state guarantees in Europe [4].

In Kazakhstan, the situation is complicated by the transitional nature of the healthcare system. On one hand, the establishment of compulsory social health insurance (CSHI) has opened new opportunities for financing expensive therapies. On the other hand, the country's geographical features, uneven regional development, and limited expert capacity create additional challenges. The opening of the Republican Center for Orphan Diseases in 2024 was an important step, but its potential remains to be realized [5].

This study does not claim to provide exhaustive coverage of all aspects of the problem. Rather, we attempted to identify the most successful international practices and assess their applicability to Kazakhstani conditions [6]. Particular attention is paid not only to formal accessibility indicators but also to institutional mechanisms that ensure system sustainability in the long term.

Objective: To compare different national approaches to ensuring orphan drug accessibility and identify directions for improving the Kazakhstani model.

2. Methodology

The study was conducted as a systematic review with elements of comparative analysis of national healthcare systems. The timeframe covers the period from 2015 to 2025, allowing consideration of current trends in orphan pharmaceutical development.

Literature Search Strategy

The search was conducted in the following databases:

- PubMed/MEDLINE (medical publications);
- Scopus (interdisciplinary research);
- Web of Science (high-impact journals);
- Cochrane Database (systematic reviews);
- Official websites of WHO, OECD, EMA, FDA.

Search queries were formulated by combining terms: "orphan drugs", "rare diseases", "pharmaceutical access", "health technology assessment", "reimbursement" with addition of specific country and region names.

The source selection process is shown in Figure 1. The final sample comprised 250 sources: peer-reviewed articles (147), regulatory documents of the Republic of Kazakhstan (43), reports from international organizations (34), and national agency documents (26).

Source Quality Assessment Criteria

For peer-reviewed publications, the following criteria were used:

- Methodological rigor and study design quality;
- Representativeness of sample/data;
- Information relevance;
- Transparency of results presentation.

For official documents, the following were assessed:

- Source authority;
- Completeness of presented data;
- Possibility of information verification.

Study Limitations

Several significant limitations of this work should be acknowledged:

Language barrier: Analysis is limited to publications in English, Russian, and Kazakh, which may have excluded important sources in other languages.

Differences in definitions: Countries use different rarity criteria for diseases, making direct comparisons difficult.

Temporal dynamics: Rapid changes in healthcare policy may render some conclusions obsolete.

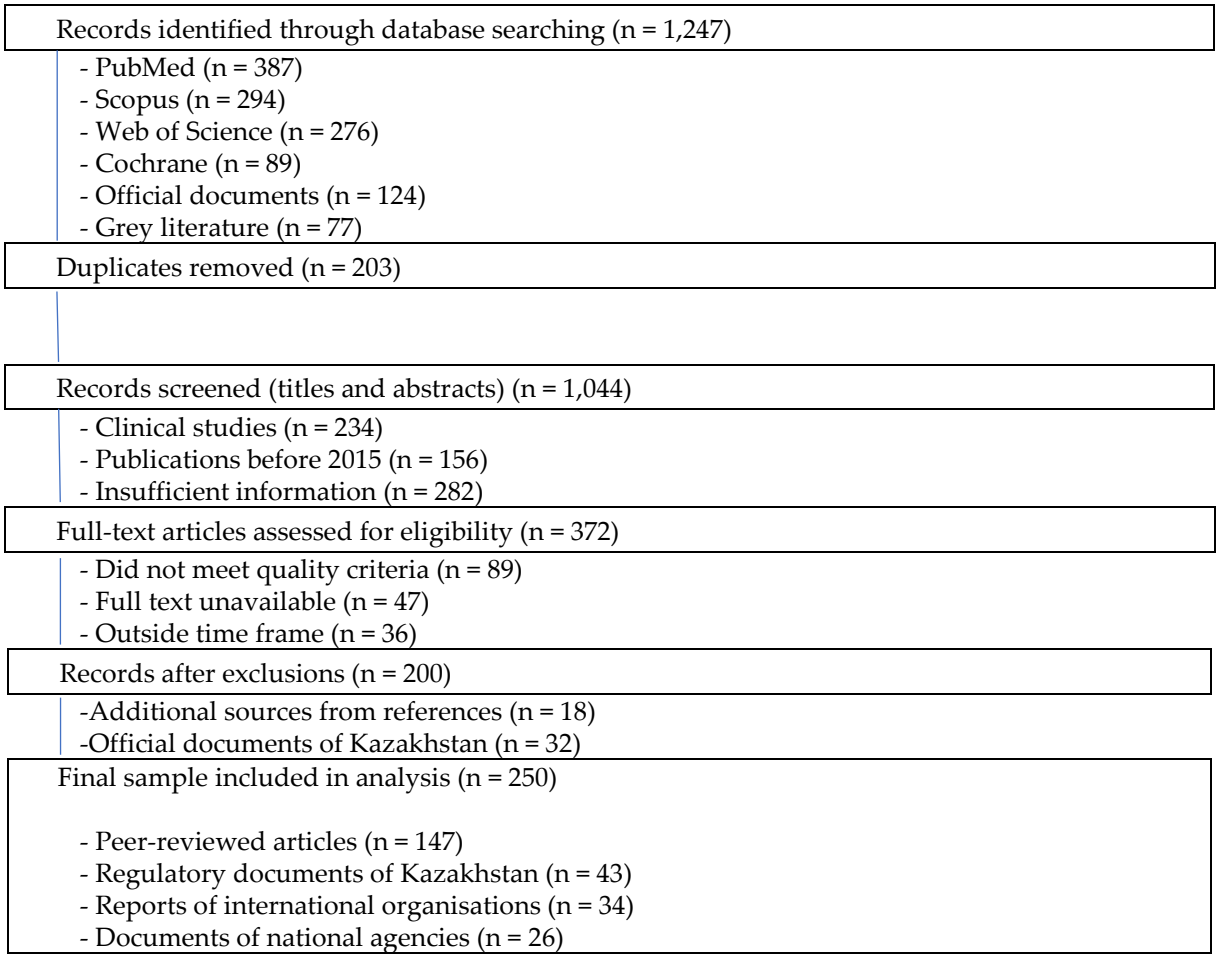
Data availability: Not all countries publish detailed statistics on orphan drugs, especially developing states.

Analysis Methods

The following analytical approaches were applied:

- Descriptive analysis of national models;
- Comparative assessment of key performance indicators;
- Multi-criteria decision analysis (MCDA) for system ranking;
- Qualitative analysis of best practices.

Performance indicators recommended by EUCERD and OECD were used to evaluate national policies in the field of rare diseases.



Source: compiled by the author

Figure 1 - PRISMA flow diagram of study selection

3. Results

Kazakhstani Model: Achievements and Gaps

Kazakhstan has made significant progress in creating an orphan drug provision system. The 2020 Code "On Public Health and Healthcare System" established legal foundations, although the definition of orphan diseases used remains less specific compared to international standards [7].

The evolution of the disease registry demonstrates gradual coverage expansion: from a modest beginning in 2015, the system has grown to 66 diseases and 205 drugs by 2024 [8]. A distinctive feature of the Kazakhstani approach is the possibility of procuring unregistered drugs—a solution dictated by the necessity to ensure access to life-saving medications in conditions of a limited local pharmaceutical market.

However, statistics reveal serious gaps. With more than 80,000 registered patients with orphan diseases, only 34,000 individuals received treatment in 2023 [9]. This means that a significant proportion of those in need remain without adequate therapy, calling into question the effectiveness of existing patient identification and routing mechanisms.

The financial burden on the system has proven substantial: over 62 billion tenge from the national budget in 2023 [10]. Meanwhile, dependence on a single distributor creates risks of supply disruptions, which have already been repeatedly observed in practice.

The establishment of the Republican Center for Orphan Diseases in June 2024 at University Medical Center was a long-awaited step toward centralizing expertise [11]. However, it is too early to judge the practical effectiveness of this initiative.

European Models: Diversity of Approaches

European experience demonstrates that no single "gold standard" exists, but some countries have achieved impressive results through various mechanisms.

Germany stands out with an 89% orphan drug accessibility rate [12]. The AMNOG system grants orphan drugs privileged status with automatic

recognition of additional benefit up to €30 million in turnover. However, critics point to the problem of "fictitious additional benefit"—of 41 drugs undergoing full assessment, most did not confirm their initial status [13].

The French approach deserves special attention due to its innovative "fixed rate" mechanism for drugs costing over €50,000 per patient annually [14]. This solution covers 80% of orphan drugs and avoids lengthy price negotiations. The HAS system ensures a median of 227 days from EMA approval to reimbursement decision—a relatively fast indicator by European standards.

The United Kingdom has pursued specialized programs, including the Highly Specialised Technologies Programme for ultra-orphan diseases [15]. A willingness to pay £100,000-300,000 per QALY appears generous, but in practice only 27 of 96 assessed orphan drugs received positive NICE recommendations.

The Dutch model of conditional reimbursement through the ODAP protocol represents an interesting compromise between accessibility and evidence [16]. The phased approach—from free provision by manufacturers to individual reimbursement upon proven effectiveness—allows collection of real-world evidence data.

American Model: Innovation and Inequality

The USA remains an undisputed leader in orphan innovations thanks to the 1983 Orphan Drug Act [17]. Seven years of market exclusivity, tax incentives, and user fee exemptions created powerful incentives for pharmaceutical companies. The result is impressive: over 650 approved orphan drugs, with approximately half of all new FDA drugs now having orphan status.

However, success in innovation has not translated into universal accessibility. The American system suffers from fragmented coverage: 85% of orphan drugs are placed on the highest tiers of Medicare Part D cost-sharing, 76% require prior authorization [18]. The 2022 Inflation Reduction Act excluded orphan drugs from Medicare price negotiations, leading to an 83% expenditure increase from 2019-2023.

Asian Systems: From Traditions to Innovations

Japan represents the most mature Asian model with universal health insurance covering the entire population [19]. The definition of orphan diseases (fewer than 50,000 patients) is more liberal compared to European standards. Special Sakigake premium bonuses for innovative drugs and 10-year exclusivity create attractive conditions for developers.

South Korea demonstrates a creative approach through risk-sharing agreements (RSA) [20]. Four types of agreements, including treatment continuation and refund mechanisms, have allowed inclusion of 88 of 156 approved orphan drugs in the reimbursement system with controlled expenditure growth to 1.44% of total pharmaceutical budget.

Singapore's public-private partnership model through the Rare Disease Fund with 3:1 co-financing represents a unique endowment approach [21]. Although coverage is limited to 8 drugs for 5 conditions, the principle of joint financing deserves attention.

Russian Experience: Systematic Approach

The Russian model warrants detailed consideration as an example of evolution from fragmented to systematic policy [22]. The high-cost nosology program has progressed from "7 nosologies" (2012-2018) to "14 nosologies" (2020-2024), covering more than 21,000 patients through centralized procurement.

The 2021 establishment of the "Circle of Good" Fund was a revolutionary decision [23]. The fund specializes exclusively in children with orphan diseases, serving 28,000 beneficiaries. Coverage of 100 diseases and 117 drugs with 413.6 billion rubles in funding since establishment demonstrates the scale of state commitments. Unique fund capabilities include procurement of unregistered drugs and rapid inclusion of new diseases.

Comparative Effectiveness Analysis

Comparison of different national models reveals several patterns that may be useful for Kazakhstan. Key indicators are shown in Table 1.

Table 1 - Comparative Characteristics of National Orphan Drug Provision Models

Country/Region	Accessibility (%)	Financing Mechanisms	Screening (number of diseases)	Key Features	Advantages	Disadvantages
Germany	89%	AMNOG, automatic recognition up to €30 million	Extended	Privileged status for orphan drugs	High accessibility, rapid inclusion	"Fictitious benefit" problem
France	78.3%	Fixed rate >€50,000/year	28	HAS median 227 days	Rapid decision-making	Limited flexibility
Netherlands	84-92%	ODAP conditional reimbursement	28	Phased approach	RWE data collection	Administrative complexity
United Kingdom	28% (27/96)	HST Programme, £100-300k/QALY	Extended	High willingness to pay threshold	Ultra-orphan consideration	Low approval rate
USA	650+ drugs	7-year exclusivity, tax incentives	Variable	Strong innovation incentives	Maximum innovations	Fragmentation, 85% with high cost-sharing
Japan	High	Universal insurance, Sakigake premium	Extended	Liberal definition (<50k)	Universal coverage	High budget burden
South Korea	56% (88/156)	RSA mechanisms, 4 types of agreements	Developing	1.44% of pharma budget	Expenditure control	Limited coverage
Russia	21,000+ patients	"Circle of Good" Fund, centralized procurement	36 (since 2023)	Children only, 100 diseases	Targeted financing	Age restriction
Kazakhstan	42% (34k/80k)	CSHI + budget, 62 billion tenge	2	66 diseases, 205 drugs	Unregistered drug procurement possible	Low patient coverage, 68 unregistered drugs

Note: *RWE - Real World Evidence; HTA - Health Technology Assessment; AMNOG - Arzneimittelmarktneuordnungsgesetz (German pharmaceutical market reorganization law); HAS - Haute Autorité de Santé (French agency); ODAP - Orphan Drugs Approval Protocol; HST - Highly Specialised Technologies; RSA - Risk-Sharing Agreements.*
Source: *Compiled by the author based on [12-23].*

First, countries with the highest accessibility rates (Germany 89%, France 78.3%, Netherlands 84-92%) share the presence of specialized orphan drug assessment procedures [24]. These procedures account for the specificity of rare diseases: small clinical trial sizes, limited evidence base, high medical need.

Second, innovative financing mechanisms are becoming the norm rather than the exception. Managed Entry Agreements (MEAs) are applied in most developed countries [25]. Italy implemented 42 agreements for 26 orphan drugs; in Catalonia, 73% of patients in PLR schemes achieved target outcomes with 11% refund to the payer.

Third, successful systems invest in creating centers of expertise and patient registries. European Reference Networks unite experts from different countries for managing complex cases [26]. Functioning registries not only help plan treatment needs but also serve as a base for post-marketing effectiveness studies.

4. Discussion

The analysis allows formulation of several important observations regarding success factors in ensuring orphan drug accessibility.

The first observation concerns the role of institutional organization. Countries with centralized decision-making systems (France, United Kingdom) demonstrate more predictable timelines for reviewing drug inclusion applications in reimbursement programs. Meanwhile, federal systems (Germany, USA) provide greater flexibility in adaptation to local conditions but at the cost of procedural complexity and potential regional inequality.

The second observation relates to the balance between innovation incentives and expenditure control. The American model maximizes incentives for new drug development but creates serious accessibility problems for patients. European systems are more effective in ensuring equal access but potentially less attractive to innovative companies. Asian countries attempt to find a middle path through risk-sharing mechanisms.

With respect to Kazakhstan, the following gaps appear most critical:

Diagnostic infrastructure. Conducting neonatal screening for only 2 diseases since 2007 appears archaic against international standards. Russia expanded screening to 36 diseases from 2023, Poland conducts screening for 28 conditions [27]. Early diagnosis is critically important for the effectiveness of many orphan disease therapies. Based on our analysis results, expanding the neonatal screening program to 15-20 diseases by 2027 following the Russian program model appears justified and achievable.

Health technology assessment procedures. The absence of specialized HTA procedures for orphan drugs leads to suboptimal decisions on drug inclusion in reimbursement lists. Standard pharmacoeconomic effectiveness criteria are often inapplicable to orphan drugs due to small patient population sizes and high clinical data uncertainty [28]. Considering European countries' experience, creating a separate unit within CSHI for orphan drug assessment with adapted criteria could improve decision-making rationale. We believe that implementing specialized HTA procedures will allow more objective assessment of the clinical and economic value of orphan drugs under conditions of limited evidence.

Regional disparities. Concentration of expertise in Almaty and Astana creates access barriers for patients from remote regions. Development of telemedicine consultations and regional expert centers could partially address this problem. In Kazakhstani conditions with its vast territory, strengthening the Republican Center for Orphan Diseases as a national expertise hub with regional branches in major cities and telemedicine connections appears a priority direction.

International integration. Kazakhstan does not yet participate in major international rare disease networks (RDI, Orphanet, European Reference Networks) [29]. For a small country, such isolation is particularly problematic as it limits access to accumulated international experience and collaborative research opportunities. Joining Orphanet, RDI, and other international rare disease networks for experience exchange and joint research could significantly strengthen the country's expert capacity.

Innovative financing mechanisms. Developed countries' experience demonstrates the effectiveness of managed entry agreements in managing uncertainty and risks. Piloting such agreements for drugs costing over 50 minimum wages per treatment course in Kazakhstani conditions could ensure balance between accessibility and financial system sustainability.

Registration barriers. The presence of 68 unregistered drugs out of 205 in the current list creates administrative complexities and procurement system risks. Simplifying registration procedures for drugs already approved by authoritative regulatory agencies (EMA, FDA) appears a logical step to improve accessibility.

At the same time, positive aspects of the Kazakhstani model should be noted. The possibility of procuring unregistered drugs demonstrates a pragmatic approach to ensuring patient access to life-saving medications. The establishment of the Republican Center for Orphan Diseases opens opportunities for systematic coordination of various stakeholder efforts.

Need for further research. Results of this review indicate the importance of conducting detailed empirical research of the Kazakhstani orphan drug provision system. In particular, analysis of real access barriers from the patient perspective, assessment of existing patient identification and routing mechanism effectiveness, and study of regional disparities in therapy accessibility are required. Such research would allow not only identification of specific problem areas of the Kazakhstani model but also development of scientifically grounded recommendations for its improvement considering local features and resource capacities.

Limitations of this study include differences in orphan disease definitions between countries, making direct comparison of accessibility indicators difficult. Additionally, rapid policy evolution in this area may render some conclusions obsolete even in the short term.

5. Conclusions

International experience in ensuring orphan drug accessibility demonstrates the absence of universal solutions but reveals several common principles of successful systems. Effective models combine specialized assessment procedures with flexible financing mechanisms, developed expert infrastructure, and active international cooperation.

Kazakhstan has good starting conditions: developed regulatory framework, significant financial resources, and political will to address the problem. The 2024 establishment of the Republican Center for Orphan Diseases creates an institutional foundation for coordinating efforts. However, achieving international quality and accessibility standards requires systemic changes in several key directions.

Comparative analysis showed that countries with the highest accessibility rates (Germany 89%, France 78.3%, Netherlands 84-92%) share the presence of adapted health technology assessment procedures for orphan drugs that account for rare disease specificity. Innovative financing mechanisms such as managed entry agreements are becoming standard practice in managing uncertainty and risks when implementing expensive orphan drugs.

For Kazakhstan, it is critically important not to mechanically copy foreign models but to adapt best practices to national characteristics: geographical conditions, demographic structure, healthcare development level, and financial capabilities. Only such an approach will help create a sustainable and effective orphan drug provision system that will serve the interests of Kazakhstani patients in the long term.

Priority development directions for the system include: expanding neonatal screening programs, implementing specialized HTA procedures, strengthening expert infrastructure through development of the Republican Center for Orphan Diseases and its regional branches, integration into international rare disease networks, and piloting innovative financing schemes with risk-sharing between the state and manufacturers.

Conflict of Interest. The author declares no conflict of interest.

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References

1. Nguengang Wakap, S., Lambert, D. M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., Murphy, D., & Rath, A. (2020). Estimating cumulative point prevalence of rare diseases: Analysis of the Orphanet database. European Journal of Human Genetics, 28(2), 165–173. https://doi.org/10.1038/s41431-019-0508-0

2. Orphanet. (2020). Prevalence and epidemiology of rare diseases: Orphanet report series. Website. [Cited 11 Sept 2025]. Available from URL: https://www.orpha.net/orphacom/cahiers/docs/GB/Epidemiology_of_rare_diseases_in_Europe.pdf

3. Chan, A. Y., Chan, V. K., Olsson, S., Fan, M., Jit, M., Gong, M., ... & Li, X. (2020). Access and unmet needs of orphan drugs in 194 countries and 6 areas: a global policy review with content analysis. Value in Health, 23(12), 1580-1591. https://doi.org/10.1016/j.jval.2020.06.020

4. Hutchings, A., Schey, C., Dutton, R., Toon, P., & Hughes, D. A. (2019). Estimating the budget impact of orphan medicines in Europe: 2010–2024. Orphanet Journal of Rare Diseases, 14(1), 1–10. https://doi.org/10.1186/s13023-019-1112-9

5. Postanovlenie Pravitel'stva Respubliki Kazakhstan. (2022). Ob utverzhdenii Kontseptsii razvitiia zdravookhraneniia Respubliki Kazakhstan do 2026 goda (Concept for the development of healthcare in the Republic of Kazakhstan until 2026) [in Russian]. Rezhim dostupa: https://adilet.zan.kz/rus/docs/P2200000945

6. OECD. (2024). OECD Health Statistics. Pharmaceutical spending and policies in OECD countries. Website. [Cited 11 Sept 2025]. Available from URL: https://www.oecd.org/health/health-data.htm

7. Kodeks Respubliki Kazakhstan. (2020). O zdorov'e naroda i sisteme zdravookhraneniia (Health of the people and the healthcare system) [in Russian]. Rezhim dostupa: https://adilet.zan.kz/rus/docs/K2000000360

8. Prikaz Ministra zdravookhraneniia Respubliki Kazakhstan. (2020). Ob utverzhdenii Pravil formirovaniia perechnia lekarstvennykh sredstv i meditsinskikh izdelii dlia besplatnogo i (ili) l'gotnogo ambulatornogo obespecheniia otdel'nykh kategorii grazhdan Respubliki Kazakhstan s opredelennymi zabolevaniiami (sostoianiiami). Utrativshii silu. (On Approval of the Rules for the Formulation of a List of Medicines and Medical Devices for Free and/or Subsidized Outpatient Care for Certain Categories of Citizens of the Republic of Kazakhstan with Certain Diseases (Conditions). Repealed) [in Russian]. Rezhim dostupa: https://adilet.zan.kz/rus/docs/V2100023783

9. Bureau of National Statistics of the Republic of Kazakhstan. (2024). Healthcare in the Republic of Kazakhstan. Statistical yearbook. Website. [Cited 11 Sept 2025]. Available from URL: https://stat.gov.kz/en/industries/social-statistics/stat-medicine

10. Social Health Insurance Fund of the Republic of Kazakhstan. (2023). Annual report for 2023. Website. [Cited 11 Sept 2025]. Available from URL: https://msqory.kz/en/eshche/o-fonde/godovye-otchety-fonda/

11. University Medical Center. (2024). Republican Centre for Orphan Diseases. Website. [Cited 14 Sept 2025]. Available from URL: https://umc.org.kz/departments/republican-center-orphan-diseases

12. Schlander, M., Garattini, S., Holm, S., Kolominsky-Rabas, P. L., Lindemann, M., O'Mahony, J. F., Paris, V., & Towse, A. (2021). Determining the value of medical technologies to treat ultra-rare disorders: A consensus statement. Journal of Market Access & Health Policy, 9(1), 1946731. https://doi.org/10.1080/20016689.2021.1946731

13. Wieseler, B., McGauran, N., & Kaiser, T. (2019). Finding studies on reimbursement of drugs for rare diseases. Bulletin of the World Health Organization, 97(4), 282–286. https://doi.org/10.2471/BLT.18.220197

14. Villa, F., Tutone, M., Altamura, G., Lazzaro, C., Trapella, G., & Garattini, S. (2019). Determinants of price negotiations for new drugs: The experience of the Italian Medicines Agency. Health Policy, 123(6), 595–600. https://doi.org/10.1016/j.healthpol.2019.03.014

15. National Institute for Health and Care Excellence. (2024). Highly Specialised Technologies Programme. Website. [Cited 14 Sept 2025]. Available from URL: https://www.nice.org.uk/what-nice-does/our-guidance/about-highly-specialised-technologies-guidanc e

16. Bouwman, L., Sepodes, B., Leufkens, H., & Torre, C. (2024). Trends in orphan medicinal products approvals in the European Union between2010–2022. Orphanet Journal of Rare Diseases, 19(1), 91. https://doi.org/10.1186/s13023-024-03095-z

17. FDA Office of Orphan Products Development. (2024). Orphan Drug Designations and Approvals Database. Website. [Cited 14 Sept 2025]. Available from URL: https://www.accessdata.fda.gov/scripts/opdlisting/oopd/

18. Bach, P. B., Giralt, S. A., & Saltz, L. B. (2017). FDA approval of tisagenlecleucel: Promise and complexities of a $450,000 cancer drug. JAMA, 318(19), 1861–1862. https://doi.org/10.1001/jama.2017.16380

19. Japanese Ministry of Health, Labour and Welfare. (2023). Pharmaceutical administration and regulations in Japan. Website. [Cited 14 Sept 2025]. Available from URL: https://www.mhlw.go.jp/english/

20. Korea Health Insurance Review & Assessment Service. (2023). National Health Insurance Statistical Yearbook 2023. Website. [Cited 14 Sept 2025]. Available from URL: https://www.hira.or.kr/bbsDummy.do?pgmid=HIRAJ030000007001

21. Singapore Ministry of Health. (2024). Rare Disease Fund: Improving access to treatment. Website. [Cited 14 Sept 2025]. Available from URL: https://www.moh.gov.sg/newsroom/rare-disease-fund/

22. Ministry of Health of the Russian Federation. (2023). Programma vysokozatratnykh nozologii: itogi 2023 goda (High-cost nosologies programme: results of 2023). [in Russian]. Website. [Cited 14 Sept 2025]. Available from URL: https://minzdrav.gov.ru/

23. Krug Dobra Foundation for Children with Severe Life-Threatening and Chronic Diseases. Godovoi otchet 2021-2024. Fond «Krug dobra» (Annual Report 2021-2024. Circle of Goodness Foundation) [in Russian]. Website. [Cited 14 Sept 2025]. Available from URL: https://static-0.minzdrav.gov.ru

24. Paulden, M., Stafinski, T., Menon, D., & McCabe, C. (2015). Value-based reimbursement decisions for orphan drugs: A scoping review and decision framework. Pharmacoeconomics, 33(3), 255–269. https://doi.org/10.1007/s40273-014-0216-7

25. Klemp, M., Frønsdal, K. B., & Facey, K. (2011). What principles should govern the use of managed entry agreements? International Journal of Technology Assessment in Health Care, 27(1), 77–83. https://doi.org/10.1017/S0266462310000803

26. European Commission. (2024). European Reference Networks. Brussels: EC. Website. [Cited 14 Sept 2025]. Available from URL: https://health.ec.europa.eu/european-reference-networks_en

27. Hatzopoulos, S., Cardinali, L., Skarżyński, P. H., & Zimatore, G. (2024). The Otoacoustic Emissions in the Universal Neonatal Hearing Screening: An Update on the European Data (2004 to 2024). Children, 11(11), 1276. https://doi.org/10.3390/children11111276

28. Iaru, I., & Vogler, S. (2024). Managed introduction of new medicines in healthcare systems. Drug Utilization Research: Methods and Applications, 233-247. https://doi.org/10.1002/9781119911685.ch21

29. Rare Diseases International. (2022). Global state of rare disease patient organizations survey 2022. Website. [Cited 14 Sept 2025]. Available from URL: https://www.rarediseasesinternational.org/wp-content/uploads/2023/09/RDI-Activity-Report-2022.pdf

30. World Health Organization. (2024). Priority diseases and reasons for inclusion. Geneva: WHO. Website. [Cited 14 Sept 2025]. Available from URL: https://www.who.int/medicines/areas/priority_medicines/Ch6_19Rare.pdf

Қазақстандағы орфандық дәрі-дәрмектердің қолжетімділігін бағалау: Мемлекеттік денсаулық сақтау жүйелерінің әртүрлі модельдерінің салыстырмалы талдауы

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Түйіндеме

Емдеудің қымбаттығына байланысты орфандық аурулар денсаулық сақтау жүйелері үшін маңызды мәселе болып қала береді. Бұл шолудың мақсаты - орфандық дәрі-дәрмектерге қолжетімділіктің халықаралық модельдерін салыстыру және Қазақстанда қолданылуы мүмкін саясат шешімдерін анықтау.

2015-2025 жылдар аралығында PubMed, Scopus және Web of Science, сондай-ақ Дүниежүзілік денсаулық сақтау ұйымы мен ЕуроОдақтың ресми құжаттарында әдебиеттерді іздеу жүргізілді. Шолу дәрі-дәрмектерге қолжетімділік, қаржыландыру механизмдері және реттеу туралы деректерді ұсынатын зерттеулерді қамтыды. Іріктеу процесі PRISMA нұсқауларына сәйкес жүргізілді.

Германия денсаулық сақтау технологияларын бағалаудың жеңілдетілген процедураларын және орфандық дәрі-дәрмектерге жеңілдіктерді автоматты түрде тануды қолданады. Франция бекітілген баға саясатын және жеделдетілген өтемақыны қолданады. АШҚ-та орфандық дәрі-дәрмектер туралы заң заманауи шешімдерді ынталандырады, бірақ теңдікте алшақтықтар сақталуда. Жапония мен Оңтүстік Корея әмбебап сақтандыру және тәуекелдерді бөлу туралы келісімдерді енгізді. Ресейде орталықтандырылған сатып алу жүйесі және «Жақсылық шеңбері» қоры бар. Қазақстанда формулярларға 205 дәрі-дәрмек кіреді, алайда неонаталдық скринингтің шектеулілігі, тіркеудің кешігуі және аймақтық айырмашылықтар секілді кедергілер әлі де бар.

Халықаралық тәжірибе денсаулық сақтау технологияларын бағалаудың мамандандырылған рәсімдерін енгізу, скринингтік бағдарламаларды кеңейту және сараптамалық орталықтарды құру әлеуетін көрсетеді. Бұл орфандық емдеуге қолжетімділікті жақсартады және денсаулық сақтау жүйесінің тұрақтылығын қамтамасыз етеді.

Түйін сөздер: орфандық аурулар, дәрі-дәрмекке қолжетімділік, денсаулық сақтау технологияларын бағалау, Қазақстан.

Оценка доступности орфанных препаратов в Казахстане: Сравнительный анализ различных моделей государственных систем здравоохранения

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Резюме

Орфанные заболевания остаются серьезной проблемой для систем здравоохранения из-за высокой стоимости лечения. Цель данного обзора — сравнить международные модели доступности орфанных препаратов и определить подходы, которые могут быть применимы в Казахстане.

Был проведен поиск литературы за 2015–2025 годы в базах данных PubMed, Scopus, Web of Science, а также в официальных документах Всемирной организации здравоохранения и ЕвроСоюза. В обзор были включены исследования, содержащие данные о доступности лекарственных препаратов, механизмах финансирования и регулировании. Процесс отбора соответствовал рекомендациям PRISMA.

В Германии используются упрощенные процедуры оценки медицинских технологий и автоматическое признание преимуществ орфанных препаратов. Во Франции применяются фиксированные тарифы и ускоренное возмещение затрат. В США Закон об орфанных препаратах стимулирует инновации, но проблемы равного доступа сохраняются. Япония и Южная Корея внедрили универсальное страхование и соглашения о разделении рисков. В России действует централизованная система закупок и фонд «Круг добра». В Казахстане в формуляры включено 205 лекарственных препаратов, однако сохраняются препятствия: ограниченный неонатальный скрининг, задержки с регистрацией и региональные различия.

Международный опыт указывает на потенциал внедрения специализированных процедур оценки медицинских технологий, расширения программ скрининга и создания экспертных центров. Это улучшит доступность лечения орфанных заболеваний и обеспечит устойчивость системы здравоохранения.

Ключевые слова: орфанные заболевания, доступность лекарственных препаратов, оценка медицинских технологий, Казахстан.