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희귀질환자 의료비지원사업 현황

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초록

희귀질환은 유병인구가 2만명 이하이거나 진단이 어려워 유병인구를 알 수 없는 질환으로 「희귀질환관리법 시행규칙 제2조」에 따라 지정 및 공고된 질환을 말한다. '희귀질환자 의료비지원사업'은 '희귀질환 산정특례'에 등록된 건강보험가입자를 대상으로 하며, 희귀질환자의 경제적 부담능력 등을 고려하여 소득재산조사를 통해 지원대상자를 선정한다. 2023년에는 소아청소년 희귀질환자에 대한 소득기준이 기준 중위소득 120% 미만에서 130% 미만으로 확대되어, 소아청소년 희귀질환자에 대한 보장성이 강화되었다. 한편, 2022년 초고가 희귀질환 치료제가 급여화 되면서 해당 치료제를 지원 받는 지방자치단체의 재정 부담이 초래되고 있는 상황이다. 또한 '희귀질환자 의료비지원사업'의 대상자 선정을 위한 소득·재산기준 및 부양의무자에 대한 소득·재산조사 기준 완화, 소득·재산조사 서류 제출 간소화 등에 대한 요구도 제기되고 있다. 질병관리청 희귀질환관리과는 향후 '희귀질환자 의료비지원사업'의 지속성 및 재정건전성을 확보하는 방안에 대해 검토하고, 소득·재산기준 및 지원체계에 대한 정비를 추진할 계획이다.

주요 검색어: 희귀질환; 의료비; 정책; 지원

서론

희귀질환은 유병인구가 2만명 이하이거나 진단이 어려워 유병인구를 알 수 없는 질환으로 「희귀질환관리법 시행규칙 제2조」에 따라 지정 및 공고된 질환을 말한다. 희귀질환은 진단이 어렵고 지속적인 치료가 필요한 경우가 많아 의료비의 부담이 과중한 질환이다. 국가와 지방자치단체는 희귀질환으로 인한 의료비 부담을 경감하고자 희귀질환자의 경제적 부담능력 등을 고려하여 2001년부터 '희귀질환자 의료비지원사업'을 통해 저소득 건강보험자에 대한 의료비를 지원하고 있다.

방법

1. '희귀질환자 의료비지원사업' 지원 개요

'희귀질환자 의료비지원사업'은 저소득 건강보험 희귀질환자를 대상으로 하며, 의료비를 지원 받기 위해서는 우선 국민건강보험공단의 '희귀질환 산정특례'에 등록되어 있어야 한

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핵심요약

① 이전에 알려진 내용은?

'희귀질환자 의료비지원사업'은 저소득 건강보험가입자에게 의료비를 지원하는 사업이며, 보건소 방문신청 또는 '희귀질환 헬프라인(http://helpline.kdca.go.kr)'을 통해 온라인으로 신청할 수 있다.

② 새로이 알게 된 내용은?

2023년에는 소아청소년 희귀질환자에 대한 소득기준이 기준 중위소득 120% 미만에서 130% 미만으로 확대되었다.

③ 시사점은?

소아청소년 희귀질환자에 대한 보장성이 강화 되었으며, 향후 의료비지원사업 소득 · 재산기준 및 지원체계에 대한 정비를 추진할 계획이다.

다. '희귀질환 산정특례' 대상질환은 전년도 신규 희귀질환으로 지정된 질환을 포함하여 결정되며, 국민건강보험공단에서 요양급여비용의 90%를 건강보험 재정으로 지원한다.

'희귀질환자 의료비지원사업'은 '희귀질환 산정특례' 대상자 중 기준 중위소득 120% 미만(2023년 부터 소아청소년 환자의 경우 기준 중위소득 130% 미만) 건강보험가입자를 대상으로 하며, '희귀질환 산정특례' 적용 후 발생하는 본인부담금 10%에 대해 추가 지원하고 있다. 또한 근육병 등 일부 중증질환 환자에게는 '보조기기 구입비, 인공호흡기 및 기침유발기 대여료, 간병비, 특수 식이 구입비' 등을 추가로 지원한다.

2. '희귀질환자 의료비지원사업' 지원절차

2022년 기준 '희귀질환자 의료비지원사업' 대상질환은 2021년에 지정 및 공고된 1,123개 희귀질환과 '희귀질환관리법 및 희귀질환자에 대한 의료비 지원기준 등에 환한 고시 경과조치'에 따른 24개 중증난치질환이다.

'희귀질환자 의료비지원사업' 지원대상자로 선정되면, 의료기관 이용 시 현장에서 진료비가 감면되며, 의료기관은 지원대상자의 진료비를 국민건강보험공단으로 청구한다. 국민건강보험공단은 시 · 군 · 구 보건소에서 전송한 대상자의 정보를 확인하고, 시 · 군 · 구 보건소가 예탁한 예탁금으로 의료기관에 진료비를 지급한다.

결과

1. 2022년 희귀질환자 분포 현황

희귀질환 목록 공고에 따라 '희귀질환자 의료비지원사업'의 대상질환은 매년 확대되고 있다[1]. 희귀질환 목록은 2018년 최초 지정 되었으며, 희귀질환으로 지정된 질환은 익년도 의료비지원 대상질환이 된다. 의료비지원 대상질환은 2019년 950개 질환, 2020년 1,038개 질환, 2021년 1,110개 질환, 2022년 1,147개 질환으로 확대되었고, 신규 희귀질환에 해당하는 지원대상자는 2019년 127명, 2020년 322명, 2021년 477명, 2022년 648명이다(표 1).

'희귀질환자 의료비지원사업'의 지역별 의료비 지급 현황을 살펴보면 경기도 21.9%, 서울 13.8%, 부산 8.4% 순이며, 수도권 환자 비율이 35.7%로 나타났다(그림 1).

연령별 지원인원이 많은 질환을 살펴보면 10대 미만은 '신생아의 호흡곤란증후군', 10대 '모야모야병', 20대 '소장 및 대장 모두의 크론병'이며, 30세 이후에서는 모두 '만성 신장병'이 지원인원이 가장 많은 질환으로 확인되었다(표 2).

표 1. 희귀질환자 의료비지원사업 대상질환 현황

연도	희귀질환	중증난치질환	의료비지원 대상질환
2018 이전	652	24	676
2019	926	24	950
2020	1,014	24	1,038
2021	1,086	24	1,110
2022	1,123	24	1,147
2023	1,165	24	1,189

단위: 개.

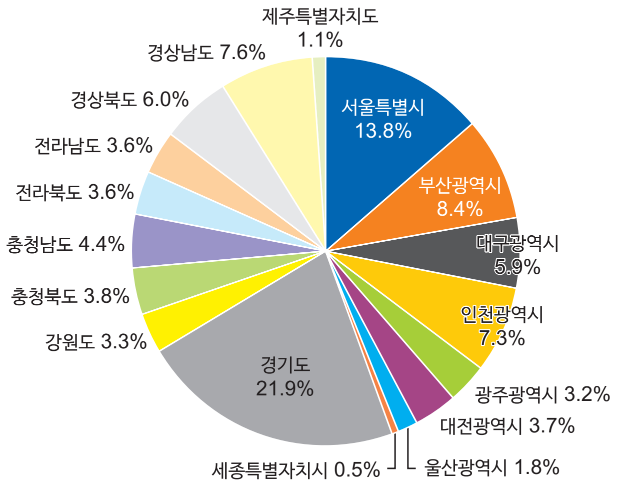

그림 1. 지역별 의료비 지급 현황 (2022년)

2. 2022년 지원항목별 의료비 지원 현황

희귀질환자가 의료기관을 이용하는 경우 ‘희귀질환 산정특례’에서 본인부담금을 우선 지원하고, 소득재산조사를 통해 일부 저소득 희귀질환자에게 본인부담금 10%에 대해 추가로 의료비를 지원하여 요양급여 중 본인부담금이 면제된다[2]. 또한 별도의 지원조건을 만족하는 경우 보조기기 구입비, 인공호흡기 및 기침유발기 대여료, 간병비, 특수조제분유 및 저단백즉석밥에 대해서도 추가로 지원하고 있다. 지원항목별 세부 지원기준은 ‘희귀질환 헬프라인 누리집(https://helpline.kdca.go.kr)’ 또는 주민등록지 관할 보건소를 통해 안내 받을 수 있다(표 3).

3. 2022년 질환별 의료비 지원 현황

지원인원이 가장 많은 질환은 ‘만성 신장병(N18)’이며, ‘크론병(K50.0–8), 모야모야병(I67.5), 유전성 제8인자결핍(D66), 강직척추염(M45.0–8)’순이다(표 4) [1].

지원금액이 가장 많은 질환은 ‘만성 신장병(N18)’이며, ‘유전성 제8인자결핍(D66), 파브리(-앤더슨)병(E75.2), 유전성 제9인자결핍(D67), A형혈우병(D66)’ 순이다(표 5).

1인당 평균 지원금액이 가장 많은 질환은 ‘헐러증후군(E76.0)’이며, ‘모르키오(-유사) (고전적) 증후군(E76.2), Ⅱ형 점액다당류증(E76.1), Ⅰ형 점액다당류증(E76.0), 헌터증후군(E76.1)’ 순이다(표 6).

1인당 지원금액이 1억 원 이상인 질환은 13개 질환이다. 이 중 혈우병 질환이 7개 질환이며, 뮤코다당증 질환이 3개 질환이다. ‘영아척수성 근위축, I형[베르드니히-호프만](G12.0)’은 2022년에 치료제가 급여화(급여비용 약 20억 원)된 영향으로 추정된다(표 7).

결론

희귀질환은 종류가 다양하고, 개별 질환에 대한 정보가 현저히 부족한 질환이다. 질병관리청 희귀질환관리과는 ‘희귀질환 헬프라인 누리집(https://helpline.kdca.go.kr)’을 통해 희귀질환에 대한 전반적 정보를 제공하고 있다. 헬프라인에서는 신규 희귀질환 지정 심의 신청 및 의료비 지원신청 등의 기능도 아울러 제공한다.

의료비지원 온라인신청 시스템은 2020년도에 최초 구축되었으며 부양의무자가 없는 환자가구에 대한 온라인신청이 우선 도입되었다. 2022년도에는 부양의무자가 있는 대상자, 소득재산조사 면제자, 정기재조사 대상자까지 온라인신청의 범위가 확대되었다. 2020년 111명, 2021년 144명, 2022년 212명이 온라인으로 의료비를 신청 하였으며, 2022년 정기재조사 대상자 71명도 온라인을 통해 정기재조사를 신청하는 등 의료비 신청의 편의성이 개선되었다.

2023년에는 소아청소년 희귀질환자에 대한 지원이 확대되었다[2]. 희귀질환은 진단이 어렵고, 지속적인 치료가 필요한 질환이며, 희귀질환의 대부분을 차지하는 유전질환은 소아청소년기에 주로 발병한다. 이에 따라 의료 취약계층인 소아청소년 희귀질환자에 대한 소득기준을 기준 중위소득 120% 미만에서 130% 미만으로 확대하여 소아청소년 희귀질환자에 대한 보장성을 강화하였다[2]. 이 밖에 정기재조사 시기에 따른 준수 기간을 명확화하여, 재조사 대상자의 서류제출 지연

표 2. 연령별 지원인원이 많은 상위 5개 질환(2022년)

연령(세)	질환명	지원인원	지원금액	1인당 평균지원금액
<10	신생아의 호흡곤란증후군	638	327	0.5
	21삼염색체증, 감수분열비분리	129	52	0.4
	A형혈우병	74	654	8.8
	담관의 폐쇄	73	115	1.6
	레녹스-가스토증후군	72	111	1.5
10–19	모야모야병	165	42	0.3
	소장 및 대장 모두의 크론병	145	96	0.7
	레녹스-가스토증후군	104	121	1.2
	유전성 제8인자결핍	97	1,138	11.7
	달리 분류되지 않은 미토콘드리아근병증	82	273	3.3
20–29	소장 및 대장 모두의 크론병	264	235	0.9
	유전성 제8인자결핍	186	2,345	12.6
	근디스트로피	156	595	3.8
	모야모야병	103	18	0.2
	대장의 크론병	90	70	0.8
30–39	만성 신장병	247	779	3.2
	유전성 제8인자결핍	206	2,893	14.0
	소장 및 대장 모두의 크론병	180	176	1.0
	기관 또는 계통 침범을 동반한 전신홍반루푸스	152	169	1.1
	소장의 크론병	107	95	0.9
40–49	만성 신장병	680	2,194	3.2
	기관 또는 계통 침범을 동반한 전신홍반루푸스	191	131	0.7
	강직척추염, 척추의 여러 부위	159	116	0.7
	유전성 제8인자결핍	159	2,127	13.4
	모야모야병	146	98	0.7
50–59	만성 신장병	1,559	5,150	3.3
	기관 또는 계통 침범을 동반한 전신홍반루푸스	158	93	0.6
	모야모야병	135	167	1.2
	근디스트로피	108	349	3.2
	베체트병	105	58	0.6
60–69	만성 신장병	2,781	9,638	3.5
	운동신경세포병	114	419	3.7
	모야모야병	108	158	1.5
	베체트병	108	59	0.5
	기관 또는 계통 침범을 동반한 전신홍반루푸스	83	58	0.7
70–79	만성 신장병	2,113	7,405	3.5
	운동신경세포병	62	215	3.5
	파킨슨병	53	81	1.5
	길랭-바레증후군	48	108	2.3
	베체트병	45	25	0.6
≥80	만성 신장병	831	2,821	3.4
	파킨슨병	29	35	1.2
	길랭-바레증후군	23	72	3.1
	특발성 폐섬유증	15	15	1.0
	노년성 황반변성(삼출성)	11	4	0.4

단위: 명, 백만 원.

표 3. 지원항목별 의료비 지원 현황(2022년)

지원항목	지원인원	지원금액
요양급여 중 본인부담금	21,209 (90.56)	71,241 (92.63)
보조기기 구입비	41 (0.18)	4 (0.01)
인공호흡기 대여료	321 (1.37)	233 (0.30)
기침유발기 대여료	197 (0.84)	37 (0.05)
간병비	1,613 (6.89)	5,324 (6.92)
특수조제분유	22 (0.09)	55 (0.07)
저단백즉석밥	18 (0.08)	19 (0.02)

단위: 명, 백만 원(%).

으로 정기재조사가 해당 반기 내 종료되지 못하는 것을 방지할 수 있도록 지침을 개정하였다[2].

'희귀질환자 의료비지원사업'은 '희귀질환 산정특례'에서 본인부담금 90%를 지원한 후 발생하는 본인부담금 10%에 대해 의료비를 지원하고 있다. 그러나 2022년에는 초고가 희귀질환 치료제가 급여화 되어, 해당 치료제를 지원 받는 지방자치단체의 재정 부담이 초래되고 있는 상황이다.

또한 '희귀질환자 의료비지원사업'의 대상자 선정을 위한

표 4. 지원인원이 많은 상위 10개 질환(2022년)

상병코드	질환명	지원인원	지원금액	1인당 평균지원금액
N18	만성 신장병	7,747	28,211	3.6
K50.0-8	크론병	1,339	1,288	1.0
I67.5	모야모야병	809	584	0.7
D66	유전성 제8인자결핍	790	10,576	13.4
M45.0-8	강직척추염	733	527	0.7
M32.1	기관 또는 계통 침범을 동반한 전신홍반루푸스	657	502	0.8
P22.0	신생아의 호흡곤란증후군	638	327	0.5
G71.0	근디스트로피	538	1,858	3.5
M35.2	베체트병	323	189	0.6
G12.2	운동신경세포병	317	1,245	3.9
그 외		8,270	31,605	3.7

단위: 명, 백만 원.

표 5. 지원금액이 많은 상위 10개 질환(2022년)

상병코드	질환명	지원인원	지원금액	1인당 평균지원금액
N18	만성 신장병	7,747	28,211	3.6
D66	유전성 제8인자결핍	790	10,576	13.4
E75.2	파브리(-앤더슨)병	182	3,557	19.5
D67	유전성 제9인자결핍	150	2,848	19.0
D66	A형혈우병	267	2,669	10.0
E76.1	Ⅱ형 점액다당류증	46	2,057	44.7
G71.0	근디스트로피	538	1,858	3.5
D67	B형혈우병	85	1,440	16.9
E75.2	고쉐병	38	1,412	37.2
G12.2	운동신경세포병	317	1,245	3.9
그 외		12,001	21,040	1.8

단위: 명, 백만 원.

표 6. 1인당 평균 지원금액이 많은 상위 10개 질환(2022년)

상병코드	질환명	지원인원	지원금액	1인당 평균지원금액
E76.0	헐러증후군	1	58	57.8
E76.2	모르키오(-유사) (고전적) 증후군	12	679	56.6
E76.1	Ⅱ형 점액다당류증	46	2,057	44.7
E76.0	Ⅰ형 점액다당류증	19	802	42.2
E76.1	헌터증후군	17	680	40.0
E76.2	마로토-라미(경도) (중증) 증후군	1	38	37.7
E75.2	고쉐병	38	1,412	37.2
E74.0	허스병	1	32	31.8
D59.5	발작성 야간혜모글로빈뇨	30	867	28.9
E75.2	니만-픽병	2	50	25.1
그 외		21,994	70,238	3.2

단위: 명, 백만 원.

표 7. 1인당 지원금액이 1억 원 이상인 질환(2022년)

상병코드	질환명	지원금액
D66	유전성 제8인자결핍	228
D66	고전적 혈우병	225
D66	유전성 제8인자결핍	187
E76.2	모르키오(-유사) (고전적) 증후군	165
E76.2	모르키오(-유사) (고전적) 증후군	150
D66	A형혈우병	144
D66	유전성 제8인자결핍	133
D66	유전성 제8인자결핍	131
D66	A형혈우병	113
G12.0	영아척수성 근위축, I형[베르드니히-호프만]	113
G12.0	영아척수성 근위축, I형[베르드니히-호프만]	111
D59.5	발작성 야간혜모글로빈뇨	102
E76.0	Ⅰ형 점액다당류증	100

단위: 백만 원.

소득·재산기준 및 부양의무자에 대한 기준 완화, 소득·재산조사 서류 제출 간소화 등에 대한 요구도 제기되고 있다. 질병관리청 희귀질환관리과는 지자체 지침 개정 교육 시 의료비지원사업 개선사항에 대한 의견을 수렴하였으며, 2023년 정책연구용역사업을 통해 '희귀질환자 의료비지원사업'의 지속성 및 재정건전성을 확보하는 방안에 대해 검토하고, 소득·재산기준 및 지원체계에 대한 정비를 추진할 계획이다.

Declarations

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National Fundholding System for Rare Disease in the Republic of Korea

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ABSTRACT

A rare disease is one for which the prevalence population is less than 20,000 or is unknown owing to difficult diagnosis. Furthermore, it refers to a disease designated and announced in accordance with Article 2 of the Enforcement Rule of the Rare Disease Control Act. The 'National Fundholding System for Rare Disease' (the National System) targets health insurance subscribers registered under the 'Special Case for Rare Disease Calculation.' Considering the economic burden of patients with rare diseases, recipients are selected through income and property investigations. The income standard for children and adolescents with rare diseases was expanded in 2023 from less than 120% of the standard median income to less than 130%. Furthermore, insurance for children and adolescents with rare diseases has been strengthened. Meanwhile, in 2022, super-expensive rare disease treatments were covered. In such situation, the financial burden of local governments receiving the support for the treatment is being caused. In addition, there is request that the standards for income and property for the selection of beneficiaries of the National System have to be eased. There is also a demand for streamlining the submission of income and property investigation documents. The Division of Rare Disease Management of the Korea Disease Control and Prevention Agency will review ways to secure the continuity and financial soundness of the National System in the future. Plans are underway to improve the income and property standards and support system.

Key words: Rare disease; Fundholding subsidy; Policy; Support

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Introduction

'Rare diseases' refer to diseases with a prevalence of less than 20,000, or with unknown prevalence due to diagnostic difficulties; designated and announced per Article 2 of the Enforcement Decree of the Rare Disease Management Act. They are difficult to diagnose and often require continuous treatment, causing a heavy financial burden. Central and local governments have, accordingly, been supporting medical costs for national health insurance beneficiaries with a low income to reduce the burden of medical expenses caused by these diseases. The 'Medical Expense Support Project for Patients with Rare Diseases' was, therefore, initiated, in consideration of the economic affordability of treatment for patients with rare

Key messages

① What is known previously?

The National System is a project to support medical expenses for low-income health insurance subscribers. You can apply by visiting a public health center or online through the 'Rare Disease Helpline (http://helpline.kdca.go.kr).'

② What new information is presented?

In 2023, the income standard for children and adolescents with rare diseases was expanded from less than 120% to less than 130% of the standard median income.

③ What are implications?

The guarantee for children and adolescent rare disease patients has been strengthened. In the future, we plan to promote the improvement of the income and property standards and support system for the National System.

diseases.

Method

1. Overview of the Support by the 'Medical Expense Support Project for Patients with Rare Diseases'

The 'Medical Expense Support Project for Patients with Rare Diseases' is for patients with rare diseases with medical expenses enrolled in the national health insurance program. In order to receive this support, they must first be registered in the 'Special Cases for Rare Disease Calculation' of the National Health Insurance Service. Diseases subject to 'Special Cases for Rare Disease Calculation' include rare diseases, newly designated in the previous year. The health insurance revenue of the National Health Insurance Corporation supports 90% of the cost of medical care benefits.

The 'Medical Expense Support Project for Patients with Rare Diseases' is targeted at health insurance beneficiaries with a median income of less than 120% (from 2023 onwards, the median income was less than 130% for pediatric and adolescent patients) among those eligible for the 'Special Cases for Rare Disease Calculation.' Additional support is provided for 10% of the out-of-pocket expenses incurred after applying the 'Special Cases for Rare Disease Calculation.' In addition, patients with serious diseases, such as muscle disorders, are provided with assistance in the 'purchase of assistive devices, the rental fee for ventilators and cough inducers, nursing expenses, and the purchase of special diets.'

2. Application Procedure for 'Medical Expense Support Project for Patients with Rare Diseases'

As of 2022, the diseases eligible for the 'Medical Expense Support Project for Patients with Rare Diseases' include 1,123 rare diseases designated and announced in 2021 and 24 serious and incurable diseases per the 'Interim Measures for Notification on the Rare Disease Management Act and Standards for Medical Expense Support for Patients with Rare Diseases.'

If a person is designated as a recipient of the 'Medical Expense Support Project for Patients with Rare Diseases,' the medical fees for his or her medical expenses are reduced. If eligible for support from the National Health Insurance Service, the recipient's information, transmitted by the associated city, county, and district public health centers, is verified by the National Health Insurance Service, and the medical expenses are paid to the relevant medical institutions, using balances deposited by the city, county, and district health centers.

Results

1. 2022 Distribution of Patients with Rare Disease

According to the announcement of the list of rare diseases, the diseases covered by the 'Medical Expense Support Project for Patients with Rare Diseases' are expanding yearly [1]. The list of rare diseases was first designated in 2018. Diseases designated as rare become eligible for medical expense support for the following year. The number of diseases covered by medical expense support has increased to 950 in 2019, 1,038 in 2020, 1,110 in 2021, and 1,147 in 2022. The number of people eligible for support projects for rare new diseases was 127 in 2019, 322 in 2020, 477 in 2021, and 648 in 2022 (Table 1).

The proportion of the medical expenses by region of the 'Medical Expense Support Project for Patients with Rare Diseases' was 21.9% for Gyeonggi-do, followed by 13.8% for Seoul, and 8.4% for Busan, indicating that the proportion of patients in the metropolitan area was 35.7% (Figure 1).

When it comes to diseases with the highest number of applicants by age, the rate of 'respiratory distress syndrome in newborns' was high for those under 10 years of age. 'Moyamoya disease' was high for adolescents; 'Crohn's disease of both the small and large intestines' was high for those in their 20s; and 'chronic kidney disease' was high for those in their 30s (Table 2).

Table 1. Status of rare diseases subject to 'National fundholding system for rare disease'

Year	Rare diseases	Intractable diseases	Status of disease
Before 2018	652	24	676
2019	926	24	950
2020	1,014	24	1,038
2021	1,086	24	1,110
2022	1,123	24	1,147
2023	1,165	24	1,189

Unit: no. of diseases.

2. 2022 Medical Expense Support by Item

When a patient with a rare disease visits a medical institution, the out-of-pocket expenses are supported by the 'Special Case for Rare Disease Calculation.' Based on an income property survey, patients with rare diseases and low income are exempted from out-of-pocket expenses, received additional support for 10% of their out-of-pocket costs [2]. In addition, if a person meets certain conditions, additional support is provided for the purchase of assistive devices; the rental fee of ventilators and cough inducers; nursing expenses; special formula; and low-protein, ready-to-eat meals. Detailed support criteria for each item can be found on the 'Rare Disease Helpline website (https://helpline.kdca.go.kr)' or obtained from the public health center under the jurisdiction of the place of residence registration (Table 3).

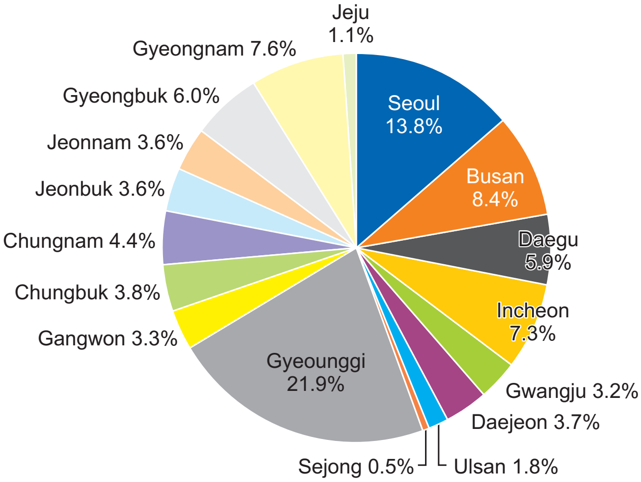

Figure 1. Regional status of 'National fundholding system for rare disease' in the Republic of Korea, 2022.

Table 2. Top 5 diseases with the highest numbers of beneficiaries by age in the Republic of Korea, 2022

Age (yr)	Diseases	Applicant	Amount	Average amount of medical expenses
<10	Respiratory distress syndrome of newborn	638	327	0.5
	Trisomy 21, meiotic nondisjunction	129	52	0.4
	Haemophilia A	74	654	8.8
	Atresia of bile ducts	73	115	1.6
	Lennox-Gastaut syndrome	72	111	1.5
10–19	Moyamoya disease	165	42	0.3
	Crohn's disease of both small and large intestine	145	96	0.7
	Lennox-Gastaut syndrome	104	121	1.2
	Hereditary factor VIII deficiency	97	1,138	11.7
	Mitochondrial myopathy, NEC	82	273	3.3
20–29	Crohn's disease of both small and large intestine	264	235	0.9
	Hereditary factor VIII deficiency	186	2,345	12.6
	Muscular dystrophy	156	595	3.8
	Moyamoya disease	103	18	0.2
	Crohn's disease of large intestine	90	70	0.8
30–39	Chronic kidney disease	247	779	3.2
	Hereditary factor VIII deficiency	206	2,893	14.0
	Crohn's disease of both small and large intestine	180	176	1.0
	Systemic lupus erythematosus with organ or system involvement	152	169	1.1
	Crohn's disease of small intestine	107	95	0.9
40–49	Chronic kidney disease	680	2,194	3.2
	Systemic lupus erythematosus with organ or system involvement	191	131	0.7
	Ankylosing spondylitis, multiple sites in spine	159	116	0.7
	Hereditary factor VIII deficiency	159	2,127	13.4
	Moyamoya disease	146	98	0.7
50–59	Chronic kidney disease	1,559	5,150	3.3
	Systemic lupus erythematosus with organ or system involvement	158	93	0.6
	Moyamoya disease	135	167	1.2
	Muscular dystrophy	108	349	3.2
	Behçet's disease	105	58	0.6
60–69	Chronic kidney disease	2,781	9,638	3.5
	Motor neuron disease	114	419	3.7
	Moyamoya disease	108	158	1.5
	Behçet's disease	108	59	0.5
	Systemic lupus erythematosus with organ or system involvement	83	58	0.7
70–79	Chronic kidney disease	2,113	7,405	3.5
	Motor neuron disease	62	215	3.5
	Parkinson's disease	53	81	1.5
	Guillain-Barré syndrome	48	108	2.3
	Behçet's disease	45	25	0.6
≥80	Chronic kidney disease	831	2,821	3.4
	Parkinson's disease	29	35	1.2
	Guillain-Barré syndrome	23	72	3.1
	Idiopathic pulmonary fibrosis	15	15	1.0
	Senile macular degeneration (exudative)	11	4	0.4

Unit: person, million won.

Table 3. Status of medical expenses by subsidy category, 2022

Subsidy	Applicant	Amount
Patient's charge in medical expenses allowance	21,209 (90.56)	71,241 (92.63)
Patient's charge in auxiliary device purchase	41 (0.18)	4 (0.01)
Patient's charge in resoiratory support device	321 (1.37)	233 (0.30)
Patient's charge in cough support device	197 (0.84)	37 (0.05)
Nursing fee	1,613 (6.89)	5,324 (6.92)
Specially formulated powdered milk	22 (0.09)	55 (0.07)
Low protein rice	18 (0.08)	19 (0.02)

Unit: person, million won.

Table 4. Top 10 diseases with the highest numbers of beneficiaries in the Republic of Korea, 2022

KCD	Diseases	Applicant	Amount	Average amount of medical expenses
N18	Chronic kidney disease	7,747	28,211	3.6
K50.0–8	Crohn's disease	1,339	1,288	1.0
I67.5	Moyamoya disease	809	584	0.7
D66	Hereditary factor VIII deficiency	790	10,576	13.4
M45.0–8	Ankylosing spondylitis	733	527	0.7
M32.1	Systemic lupus erythematosus with organ or system involvement	657	502	0.8
P22.0	Respiratory distress syndrome of newborn	638	327	0.5
G71.0	Muscular dystrophy	538	1,858	3.5
M35.2	Behçet's disease	323	189	0.6
G12.2	Motor neuron disease	317	1,245	3.9
Others		8,270	31,605	3.7

Unit: person, million won. KCD=Korean standard classification of diseases.

3. 2022 Status of Medical Expense Support by Disease

The disease with the highest number of applicants was 'chronic kidney disease (N18)', followed by 'Crohn's disease (K50.0–8); Moyamoya disease (I67.5); hereditary factor VIII deficiency (D66); and ankylosing spondylitis (M45.0–8)' (Table 4) [1].

The disease with the highest amount of required balance support was 'chronic kidney disease (N18)', followed by 'hereditary factor VIII deficiency (D66); Fabry's (-Anderson) disease (E75.2); hereditary factor IX deficiency (D67); and hemophilia A (D66)' (Table 5).

The disease with the highest average amount of required balance support per person was 'Heller's syndrome (E76.0)'; followed by 'Morquio (-like) (classic) syndrome (E76.2); Mucopolysaccharidosis, type II (E76.1); type I mucopolysaccharides (E76.0); and Hunter syndrome (E76.1)' (Table 6).

There were 13 diseases for which the required balance support per person was more than 100 million won. Among them, hemophilia diseases numbered seven and mucopolysaccharidosis diseases amounted to three. It is estimated that costs were affected by the inclusion of individuals with 'Infantile spinal muscular atrophy, type I [Berdnig-Hoffmann] (G12.0),' who were reimbursed for their treatment in 2022 (about 2

Table 5. Top 10 diseases with the highest amount of medical expenses in the Republic of Korea, 2022

KCD	Diseases	Applicant	Amount	Average amount of medical expenses
N18	Chronic kidney disease	7,747	28,211	3.6
D66	Hereditary factor VIII deficiency	790	10,576	13.4
E75.2	Fabry's (-Anderson) disease	182	3,557	19.5
D67	Hereditary factor IX deficiency	150	2,848	19.0
D66	Haemophilia A	267	2,669	10.0
E76.1	Mucopolysaccharidosis, type II	46	2,057	44.7
G71.0	Muscular dystrophy	538	1,858	3.5
D67	Haemophilia B	85	1,440	16.9
E75.2	Gaucher's disease	38	1,412	37.2
G12.2	Motor neuron disease	317	1,245	3.9
Others		12,001	21,040	1.8

Unit: person, million won. KCD=Korean standard classification of diseases.

Table 6. Top 10 diseases with the highest average amount of medical expenses in the Republic of Korea, 2022

KCD	Diseases	Applicant	Amount	Average amount of medical expenses
E76.0	Hurler syndrome	1	58	57.8
E76.2	Morquio (-like) (classic) syndrome	12	679	56.6
E76.1	Mucopolysaccharidosis, type II	46	2,057	44.7
E76.0	Mucopolysaccharidosis, type I	19	802	42.2
E76.1	Hunter's syndrome	17	680	40.0
E76.2	Maroteaux-Lamy (mild) (severe) syndrome	1	38	37.7
E75.2	Gaucher's disease	38	1,412	37.2
E74.0	Hers' disease	1	32	31.8
D59.5	Paroxysmal nocturnal haemoglobinuria [Marchiafava-Micheli]	30	867	28.9
E75.2	Niemann-Pick's disease	2	50	25.1
Others		21,994	70,238	3.2

Unit: person, million won. KCD=Korean standard classification of diseases.

billion won in reimbursement costs) (Table 7).

Conclusion

There are various types of rare diseases, and information about individual diseases is significantly lacking. The Rare Disease Management Division of the Korea Disease Control and Prevention Agency provides general information on rare diseases through the 'Rare Disease Helpline (https://helpline.kdca.go.kr).' On the Helpline, assistance with the application of a rare new disease designation review and application for medical expense support is provided.

The online application system for medical expense support was first established in 2020, and online application was first introduced for patient households without dependents. In 2022, the scope of online applications expanded to include

Table 7. Top 10 diseases with amount per person exceeds 100 million won in the Republic of Korea, 2022

KCD	Diseases	Amount
D66	Hereditary factor VIII deficiency	228
D66	Classical haemophilia	225
D66	Hereditary factor VIII deficiency	187
E76.2	Morquio (-like) (classic) syndrome	165
E76.2	Morquio (-like) (classic) syndrome	150
D66	Haemophilia A	144
D66	Hereditary factor VIII deficiency	133
D66	Hereditary factor VIII deficiency	131
D66	Haemophilia A	113
G12.0	Infantile spinal muscular atrophy, type I [Werdnig-Hoffman]	113
G12.0	Infantile spinal muscular atrophy, type I [Werdnig-Hoffman]	111
D59.5	Paroxysmal nocturnal haemoglobinuria [Marchiafava-Micheli]	102
E76.0	Mucopolysaccharidosis, type I	100

Unit : million won. KCD=Korean standard classification of diseases.

those with dependents, those exempt from income and property inspections, and those subject to periodic re-inspections. In 2020, 111 people, 144 in 2021, and 212 in 2022, applied for medical expense support online, and 71 people who were subject to regular re-surveys in 2022 also applied for periodic re-surveys online, benefitting from the convenience of the application procedure.

In 2023, the support for pediatric and adolescent patients with rare diseases expanded [2]. Rare diseases are difficult to diagnose and require continuous treatment. Genetic diseases account for most rare diseases in childhood and adolescence. Accordingly, the income criteria for pediatric and adolescent patients with rare diseases, both medically vulnerable groups, were expanded from less than 120% to less than 130% of the median income to strengthen coverage for children and adolescents with rare diseases [2]. In addition, the guidelines have been revised to clarify the compliance period according to the timing of the periodic re-survey and to prevent periodic re-inspections from being completed within the relevant half-year with a delay in the submission of documents by the person subject to the re-survey [2].

The 'Medical Expense Support Project for Patients with Rare Diseases' covers medical expenses for 10% of the out-of-pocket expenses incurred after 90% of the out-of-pocket expenses of the 'Special Case for Rare Disease Calculation' has been paid. In 2022, however, rare disease treatment drugs with very high prices were subsidized, causing a financial burden on local governments.

In addition, there are demands that the income and property standards used to select recipients for the 'Medical Expense Support Project for Patients with Rare Diseases' be eased and that the submission of income and asset investigation documents be simplified. The Rare Disease Management Division of the Korea Disease Control and Prevention Agency collected opinions relating to the improvement of the medical expense support project during the revised local government guidelines training. Through the 2023 policy research project, we plan to examine ways of ensuring the sustainability and financial soundness of the 'Medical Expense Support Project for Patients with Rare Diseases' and to promote improved income property standards and support systems.

Declarations

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