The usage of enzyme replacement treatments, economic burden, and quality of life of patients with four lysosomal storage diseases in Shanghai, China

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Abstract

Background

Lysosomal storage diseases (LSDs) are a group of rare diseases that caused progressive physical dysfunction and organ failure, which significantly affected patients' quality of life. Enzyme replacement treatments (ERTs) are now acknowledged as the advanced therapies for LSDs while cost millions per patient per year. Previous studies seldom reported the usage of ERTs and disease burden of patients with LSDs in China. The objective of this study was to explore the characteristics and usage of ERTs of patients with the four different LSDs (Gaucher, Fabry, Pompe disease and Mucopolysaccharidosis) in Shanghai and then evaluate the economic burden and quality of life of these patients.

Methods

The study used data extracted from a large survey of living conditions of patients with rare diseases in Shanghai, which was conducted from April to August 2020. A total of 31 patients, involving 5, 14, 4 and 8 patients with Gaucher, Fabry, Pompe disease and Mucopolysaccharidosis, respectively, was included in analysis. Descriptive statistics was used to describe the socio-demographic information (age, gender, education and etc.), economic burden caused by LSDs (direct medical and non-medical costs, and indirect cost in 2019), the treatment (usage of drugs) and the patients' quality of life.

Results

Five Gaucher disease patients in Shanghai used Imiglucerase in 2019, while the other 26 patients with the other three LSDs didn't receive ERTs. The total health expenditure of Gaucher disease patients was 2,273,000 CNY on average mainly resulted by the high cost of Imiglucerase. The total health expenditure of the other 26 patients was 37,765 CNY on average. The average total disease burdens of Gaucher disease patients and the patients with the other three LSDs were 164,301 CNY and 58,352 CNY, respectively. The mean EQ-VAS score of GD patients was 76.4 ± 15.5 , which was higher than that of the other three LSDs. All the patients with LSDs in this study reported poor quality of life, which was significantly worse than the Chinese general population.

Conclusion

Few patients with LSDs in Shanghai could have access to available ERTs without a high reimbursement level. Though the cost-sharing mechanism of basic medical insurance, charity fund and patients had been explored for Gaucher disease in Shanghai, the Out-of-pocket part still laid a heavy economic burden on the patients and their families. The scope of drug reimbursement list and the reimbursement level should be further expanded and raised to help improve the quality of life of patients with LSDs.

Introduction

Lysosomal storage diseases (LSDs) are a group of diseases caused by defects in single genes. Enzyme defects cause nearly seventy percent of the LSDs, and the rest are defects in enzyme activator or associated proteins (1). A deficit in any of these enzymes will result in progressive accumulation of materials in affected organs and tissues, which will result in an increase in the size and number of these organelles and finally in cellular dysfunction and organ failure (2). Though as a group, LSDs are with an estimated incidence of 1/5,000 to 1/5,500 (2), individual LSDs are usually recognized as rare diseases with estimated incidences ranging from 1/50,000 to 1/250,000 live births (3). There have been seventy LSDs reported so far, while only 7 of them have approved therapies on the global market (4), most therapies are enzyme replacement treatments (ERTs). Though there are approved treatments for the 7 LSDs, but the cost for these treatments is extremely high (5)(6). Some of the Western countries have already published policies regarding rare diseases (7)(8), and a few have further designed reimbursement measures for patients with LSDs with high costs. For example, the National Institute for Clinical Excellence raised the concept of ultra-orphan drugs (9) and designed the highly specialized technology (HST) appraisal process to recommend on the use of new and existing highly specialized medicines and treatments, which are usually very expensive within the National Health Service (NHS) in England. The HST guidance has recommended the use of Eliglustat and Migalastat for Gaucher disease (GD) type 1 and Fabry disease (FD), respectively (10)(11). The Australian government initiated the Life Saving Drugs Program in 1994 to reimburse expensive and life-saving drugs for life threatening and rare diseases. Now, the program provides fully subsidized access to 16 drugs for patients with 10 indications, including GD, FD, Pompe Disease (PD), Mucopolysaccharidosis (MPS) type I, type II, type IVA, type VI and Neuronal ceroid lipofuscinosis type 2 (CLN2), which are LSDs (12).

With the development of socio-economic and universal health coverage in China (13), the attention on rare disease has turned from the provincial and city level to the national level. The first batch of National Rare Diseases List (NRDL) was published in 2018 (14). After that, the Chinese government has published a series of policies to improve comprehensive healthcare security of rare diseases, including constructing the diagnosis and treatment network, registering rare disease patients, publishing guidelines for diagnosis and treatment, improving the accessibility, reducing the import tax, accelerating the market authorization process of drugs for rare diseases, and bring the drugs for rare diseases into the National Drug Reimbursement List (NDRL) (15).

There are 16 different approved therapies for 7 LSDs in the world (4), while there are only altogether 8 available therapies in China for 5 LSDs, which are GD, FD, MPS, PD, and Niemann-Pick disease (NP), in the NRDL. However, only Miglustat for NP type C is now included in the NDRL, while the other seven ERTs are not. See Table 1 for the details. Recently, the National Healthcare Security Administration claimed that it has basically included all the drugs for rare diseases meeting certain criteria and could not include some drugs into the NRDL due to the extremely high cost, which is far beyond the payment capacity of the basic medical insurance fund and the patients (16). The newly updated NDRL (2020 version, published on Dec 28, 2020) did not contain these extremely expensive

drugs for rare diseases(17). Theoretically, there are no healthcare security measures on the national level in China for patients with the mentioned four LSDs, which are GD, PD, FD and MPS.

Table 1 Marketed and reimbursed drugs for LSDs in China

NRDL code	Disease	Approved name	Brand name	Approved date in China	Included in NDRL
27	Fabry disease	Agalsidase beta ^a	Fabrazyme	2009/12	No
31	Gaucher disease	Imiglucerase	Cerezyme	2008/11	No
		Velaglucerase ^a	Vpriv	No	—
		Taliglucerase ^a	Elelyso	No	—
		Miglusta	Zavesca	No	—
		Eliglustat ^a	Cerdelga	No	—
35	Pompe disease	Alglucosidas alfa	Myozyme	2017/12	No
		Agalsidase alfa	Replagal	2020/8	No
		Migalastat ^a	Galafpld	No	—
73	MPS				
	Type I	Laronidase ^a	Aldurazyme	2020/6	No
	Type II	Idursulfase ^a	Elaprase	2020/9	No
	Type IVA	Elosulfase ^a	Vimizim	2019/6	No
	Type VI	Galsulfase	Naglazyme	No	—
82	Niemann–Pick disease type C	Miglustat	Zavesca	2017/9	Yes
	Wolman disease	Sebelipase	Kanuma	No	—
	Neuronal ceroid lipofuscinosis type 2(CLN2)	Cerliponase	Brineura	No	—

a: Drugs included in the List of Urgently Needed New Drugs from Overseas for Clinical Use.

Current studies regarding LSDs patients in China are mainly from the clinical aspect, while few are not. *Chen et al.* introduced the demographic characteristics and distribution of all 322 diagnosed patients LSDs in Eastern China, including Shanghai and other six provinces(18). *Zhao et al.* studied the characteristics of 59 Chinese PD patients from the Pompe Registry(19). *Yang et al.* described the cost-sharing mechanism for Imiglucerase in Qingdao, Shandong province(20). Except for the mentioned three literatures, there are some large-scale surveys focusing on living conditions of patients with rare disease in China. Some surveys on LSDs did report the cost of illness while the usage of ERTs remained unknown. (21)(22) (23) (24). Furthermore, some of these reports are not able to access the full texts.

Shanghai, as one of the most developed cities in China, has over 242 million residents, with an average GDP of 134,982 CNY(25). It is one of the first cities that reimbursed Imiglucerase for GD patients. In 2011, Shanghai Children's Hospitalization Assistance Fund, managed by the Red Cross Society of China Shanghai Branch decided to reimburse the ERTs for patients with the mentioned four LSDs, with a maximum reimbursement amount of 100,000CNY per patient per year (26). In 2013, Imiglucerase could be paid by the basic medical insurance in Shanghai and reimbursement level was ranging from 80%-85% depending on the dosage. In 2017, the Shanghai Foundation for Rare Disease established a special assistance fund for LSD patients supported by enterprises(27). The assistant amount was decided based on the income level of patients, ranging from 70% to 100% of the Out-of-Pocket (OOP) part, who were receiving ERTs treatments. The outcomes of these policy intervention on patients with LSDs in Shanghai are little unknown except that *Cai et al.* reported the numbers of inpatient and outpatient visits of GD and FD patients in Shanghai, but the direct medical costs of these patients were not reported separately(28). Our study aimed to explore the characteristic and usage of ERTs of patients with GD, PD, FD and MPS in Shanghai and then evaluated the economic burden and quality of life (QoL) of these patients.

Methods

Study design

This study focusing on patients with 4 LSDs was based on a large survey of living conditions of patients with rare diseases in Shanghai. The survey used a self-designed questionnaire based on a literature review and interviews with several doctors, health economists and government officials in the field of rare diseases. Snowballing sampling method was adopted due to that there was no real epidemiological data nor patient registry of patients with rare diseases in Shanghai. Due to COVID-19, face-to-face survey was cancelled. The electronic questionnaire was administered using the Wenjuanxing software (Changsha Ranxing Information Technology Co. LTD) and filled out online by patients with rare diseases or their primary caregivers. The participants recruitment process was conducted through online and offline platforms. The doctors in hospitals, which act as members of the National Rare Disease Diagnosis and Treatment

Network, in Shanghai helped to invite their diagnosed patients with rare diseases to participate in the survey. Several patient organizations of rare diseases also called on patients to involve in the investigation. The whole process of data collection was done from April to August 2020.

Inclusion criteria

The patients would only be included in the survey when they met the following conditions:

- a. The disease they were diagnosed with were recognized as rare disease on the list of NRDL or Orphanet(29).
- b. They paid their Basic Medical Insurance premium in Shanghai, including urban employee and urban resident basic medical insurance.
- c. The patients or their primary caregivers were willing to participate in the survey and were able to complete the online questionnaire.

Quality control

The follow-up telephone calls of each participant were made to ensure the quality of data. The follow-up could correct the obvious mistakes participants made and refill in the blanks they left. The follow-up visits were performed by four postgraduates major in health policy or health management research, who had been strictly and systematically trained before.

Data extraction

The information of patients diagnosed with GD, PD, FD and MPS were extracted from the dataset. A total of 31 patients were enrolled, including 5 GD patients, 4 PD patients, 14 FD patients and 8 MPS patients, in this study. 18 patients answered the questionnaire themselves and the rest 13 patients’ conditions were reported by their caregivers due to reasons like “the patient cannot read”.

Several important variables were chosen from the long questionnaire, including socio-demographic information (birthday, gender, education, marriage, occupation, personal annual income, annual household income, and etc.), economic burden caused by the disease (the components of direct medical costs, direct non-medical costs and indirect cost in 2019), the treatment (usage of drugs) and health states (quality of life measured by EQ-5D-Y-3L and EQ-5D-3L for different age groups).

Data analysis

Since the included participants were too little (n=31), mainly descriptive statistics were used for the analysis. The EQ-5D Visual Analogue Scale (VAS) scores and problems reported in 5 dimensions (mobility, self-care, usual activities, pain/discomfort, and anxiety/depression) were used to evaluate the patients’ QoL. One-way ANOVA analysis and Fisher exact test were used to test whether the QoL of patients with each LSD was statistically different. Statistical analysis was performed using SPSS 20.0 software. *P* value less than 0.05 was considered significant.

Ethical statements

Informed consents were attained by all the participants before the formal survey started. The participants’ privacy, including any individual information they provided in the survey, would be protected. This study was approved by the Medical Ethics Committee of Shanghai Health Development Research Center (No. 2020004).

Results

General characteristics

Table 2 demonstrates the characteristics of patients with 4 LSDs in Shanghai. Seventeen of the included patients were male (54.8%). Ten were non-adult among the 31 patients and eight of them were boys (80.0%). The mean age of the sample was 29.8±14.4 years. Five patients failed to complete their education as they suffered from the diseases. Only 12 of the 21 adult patients (57.1%) were employed in 2019. Thirteen of the 21 adult participants were married. Only 1 of the married adults hadn’t given birth to a child. Altogether 16 participants had urban resident basic medical insurance and 15 participants had urban employee basic medical insurance. Additionally, two patients purchased commercial health insurances. The mean personal and household annual incomes were 57,218CNY and 184,987CNY (1USD≈6.8CNY), respectively.

Table 2 The socio-demographic characteristics of patients with LSDs

Characteristics	Overall (N=31)		Non-adult (N=10)		Adult (N=21)	
	N	%	N	%	N	%
Gender						
Male	17	54.8	8	80.0	9	42.9
Female	14	45.2	2	20.0	12	57.1
Mean age ($\bar{x} \pm S$)	29.8±14.4		12.4±3.3		38.1±9.5	
Educational level						
No education	4	12.9	4	40.0	0	0.0
Primary school	2	6.5	1	10.0	1	5.0
Middle school	7	22.6	5	50.0	2	9.5
High school	4	12.9	0	0.0	6	28.6
College or higher	14	45.2	0	0.0	12	57.1
Employment status						
Employed	—	—	—	—	12	57.1
Unemployed	—	—	—	—	8	38.1
Retired	—	—	—	—	1	4.8
Marriage						
Married	—	—	—	—	13	61.9
Single	—	—	—	—	6	28.6
Divorced	—	—	—	—	2	9.5
Fertilityn						
No	—	—	—	—	9	42.9
Yes	—	—	—	—	12	57.1
Medical Insurance						
Urban Employee Basic Medical Insurance	15	48.4	0	0.0	15	71.4
Urban Resident Basic Medical Insurance	16	51.6	10	100.0	6	28.6
Additional commercial insurance	2	6.5	0	0.0	2	9.5
Personal income per year(CNY, 1USD≈6.8CNY)						
0	—	—	—	—	4	19.0
10,000-49999	—	—	—	—	5	23.8
50000-99999	—	—	—	—	2	9.5
100000-199999	—	—	—	—	8	38.1
200000-299999	—	—	—	—	2	9.5
Household annual income (CNY, 1USD≈6.8CNY)						
10000-49999	1	3.2	1	10.0	0	0.0
50000-99999	6	19.4	1	10.0	5	23.8
100000-199999	9	29.0	4	40.0	5	23.8
200000-299999	12	38.7	4	40.0	8	38.1
Above 300000	3	9.7	0	0.0	3	14.2

Usage of ERTs

Among all the participants, all the 5 GD patients were using ERTs in 2019, while none of the rest 26 were treated with ERTs. The main reason why they never used ERTs is that “the treatment is too expensive to afford”. Additionally, one replied with “Has just been diagnosed with the disease, has not started treatment” and one replied with “Worried about the adverse events”.

Health expenditure

The mean total health expenditure of patients with GD was 2,273,100CNY in 2019 while that of patients with the other 3 LSDs (PD, FD and MPS) was 37,765CNY. The higher percent of OOP cost in outpatient expenditure among GD patients was caused by one GD patient who received a spine surgery and was followed up in outpatient. The inpatient expenditure contributed 2,234,400 CNY, which was over 98.3% of the total cost of GD. Notably, the cost for Imiglucerase accounted for 99.9% of the total inpatient cost, and of which 79.0% could be covered by basic medical insurance and 15.6% was funded by Shanghai Foundation for Rare Disease. The GD patients also needed to pay the rest 5.4% by themselves, which was still 121,200 CNY in average. The medical cost of the other 3 LSDs, including 68.9% outpatient cost and 13.4% inpatient cost could be covered by basic medical insurance. The mean out-of-pocket expenditure for patients with the other 3 LSDs was 21,367CNY in total. The mean cost happened outside of hospital (which includes cost for drugs and medical devices purchased from retail pharmacies) was similar for GD patients (4,300CNY) and patients with the other 3 LSDs (4,700CNY). However, expenditure happened outside of hospital should be self-paid. See Table 3 for details.

Table 3 Health expenditure of patients with LSDs in 2019 (N=31)

Cost (CNY)	GD(N=5)		PD,FD and MPS(N=26)	
	Mean	Median	Mean	Median
Total	2273000	1648000	37765	4700
Outpatient				
Total	34400	4000	13088	2450
Basic medical insurance	300	0	9017	1400
Out-of-pocket	34100	2500	4071	750
Inpatient				
Total	2234400	1612000	19977	0
Basic medical insurance	1764244	0	2681	0
Charity	348956	262080	0	0
Out-of-pocket	121200	100000	17296	0
Outside the hospital	4300	0	4700	1000

Economic burden of patients

The average economic burden of patients caused by GD was 164,301CNY, while the average economic burden of patients with PD, FD and MPS was 58,352CNY in 2019. Direct medical cost was the majority of the disease burden, which took 97.1% and 60.5% for GD patients and patients with the other three LSDs, respectively. The indirect cost of patients with PD, FD and MPS was 21,860CNY, which was higher than that of GD patients.

Table 4 Economic burden of patients with LSDs in 2019 (N=31)

Cost (CNY)	GD(N=5)			PD, FD and MPS(N=26)		
	Mean	Median	%	Mean	Median	%
Total economic burden	164301	112500	100.0	58352	7000	100.0
Direct medical cost	159600	102500	97.1	35321	4000	60.5
Direct non-medical cost	2361	2000	1.4	1171	50	2.0
Indirect cost	2340	2200	1.4	21860	0	37.5

Quality of life

The QoL of patients with individual LSD was shown in Table 5. The mean EQ-VAS scores of patients with GD, FD, PD and MPS were 76.4, 55.0, 52.0, and 46.0, respectively. The mean EQ-VAS score of GD patients was the highest, but there was no significant difference among the QoL of patients with 4 different LSDs ($P>0.05$). The majority of patients with LSDs reported problems in Pain/discomfort and Anxiety/depression dimensions, accounts for 80.6% and 74.2%. All the GD patients reported no problems in Mobility, while 35.7%, 100% and 75% of patients with FD, PD, and MPS reported problems. The difference was statistically significant ($\chi^2=11.542$, $P<0.01$). Significant difference was also found in Self-care dimension among the 4 LSDs ($\chi^2=16.679$, $P<0.001$), and GD patients had better performance than patients with PD and MPS.

Table 5 The QoL of patients with LSDs in 2019 (N=31)

EQ-5D-3L Dimension	Problems	Total n(%)	Treated with ERT(N=5) GD[n(%)]	Not treated with ERTs(N=26)			χ^2 F	P
				FD[n(%)]	PD[n(%)]	MPS [n(%)]		
EQ-VAS(Mean±SD)		31(100)	76.4±15.5	55.0±19.7	52.0±12.9	46.0±28.6	2.171	0.115
Mobility	No	16(51.6)	5(100)	9(64.3)	0(0)	2(25.0)	11.542	0.004*
	Yes	15(48.4)	0(0)	5(35.7)	4(100)	6(75.0)		
Self-care	No	23(74.2)	5(100)	14(100)	2(50.0)	2(25.0)	16.679	<0.001**
	Yes	8(25.8)	0(0)	0(0)	2(50.0)	6(75.0)		
Usual activities	No	13(41.9)	4(80.0)	7(50.0)	1(25.0)	1(12.5)	6.301	0.076
	Yes	18(58.1)	1(20.0)	7(50.0)	3(75.0)	7(87.5)		
Pain/discomfort	No	6(19.4)	2(40.0)	1(7.1)	1(25.0)	2(25.0)	3.477	0.285
	Yes	25(80.6)	3(60.0)	13(92.9)	3(75.0)	6(75.0)		
Anxiety/depression	No	8(25.8)	3(60.0)	2(14.3)	1(25.0)	2(25.0)	3.863	0.266
	Yes	23(74.2)	2(40.0)	12(85.7)	3(75.0)	6(75.0)		

*p<0.05,**p<0.01

Discussion

This is the first study focusing on the usage of all the available ERTs for LSDs in China, as well as the disease burden and QoL of patients with GD, FD, PD and MPS as well.

The results showed that the patients using ERTs in Shanghai were still the minority. Only 16.1% (5/31) of the participants, which were 5 GD patients. The average total health expenditure of GD patients was significantly higher than that of the other patients in this study, resulted by the high cost of Imiglucerase. The outpatient cost of GD patients was higher than that of the others as well. The reason was that one of the GD patients received spine surgery and had multimorbidity. The average outpatient expenditure would be 13,000CNY if the patient were excluded from the analysis. The inpatient cost of GD patients was extremely high, contributed by the cost of Imiglucerase, which needed to be intravenous injected through hospitalization procedure. The health expenditures of the patients with the other LSDs were caused by different support treatment.

As there were 5 GD patient receiving ERTs in 2019, which indicated that the reimbursement level directly determined whether the patients with LSDs could afford the ERTs. Though the basic medical insurance in Shanghai started to reimburse Imiglucerase from 2013, based on our interview with the 5 GD patients, none of them started to use it until the Shanghai Foundation for Rare Disease established the special assistance fund in 2017. The reason was that, unlike common drugs, the OOP part after reimbursement was still unaffordable to the patients, which was estimated to be 300,000-400,000CNY per patient per year. The Shanghai Foundation for Rare Disease reimbursed the patients depending on their personal income levels, which meant the lower their personal income is, the more reimbursement they would get. For this cost-sharing mechanism, the basic medical insurance played major role, which covered the majority of expenditures, while the assistance fund played the supplementary role. The patients with LSDs in few Chinese provinces or cities who have access to the ERTs rely on the high reimbursement levels as well. The Qingdao government, in Shandong province, established a supplementary medical insurance to cover 80% of the cost for Imiglucerase. The donation from the enterprises and civil assistance for low-income families would cover some of the rest part as well(30). The Zhejiang government settled a special fund for rare diseases in 2020, mainly focusing on the expensive drugs for rare diseases. The fund reimburses has included three drugs for LSDs, which are Imiglucerase, Alglucosidas alfa for PD, and Agalsidase beta for FD, respectively(31).

Though the cost for Imiglucerase was mostly covered by basic medical insurance and special assistance fund in Shanghai, the OOP expenditure of GD patients was still high, which was approximately 121,200CNY in 2019, contributing to a major part of the disease burden. The direct medical cost was almost 2 times of the disposable income per capital in Shanghai, which was 69,442CNY in 2019(32). The 8 GD patients' average OOP expenditure for Imiglucerase was 82,700CNY in Qingdao in 2017(20), while the ceiling of OOP is 100,000 per patient per year in Zhejiang(33). In addition, the direct non-medical costs of GD patients in Shanghai were still higher than that of the patients with other 3 LSDs. The reason was that the GD patients needed to go to the hospital for treatment every two weeks.

As LSDs usually cause progressive damage in connective tissue, skeletal structure and various organs(34), pain and physical discomfort were the most frequently mentioned symptoms by patients, which was also reported in our study. The EQ-VAS scores of patients with all the four included LSDs were lower than the Chinese population norm of 80.4,(35) revealing the impaired QoL in patients with LSDs. Among the 4 LSDs, GD patients had highest mean EQ-VAS score (76.4), which was quite close to the norm. The mobility and self-care ability of GD patients were significantly better than patients with FD, PD and MPS as well. The findings supported that receiving ERTs is meaningful to the patients and could improve their QoL, which was also reported in the previous study(36). Furthermore, with the better health status, eighty percent of the GD patients worked as normal people did, while only 50% (8/16) of the adult patients with the other three LSDs could go to work. Most GD patients didn't need others to take care of them, which may be the reason why the indirect cost of GD patients (2,340CNY) was much lower than that of the patients with the other three LSDs (21,860CNY).

Our study also has some limitations. Based on the interviews with some doctors and key opinion leaders from some rare disease organizations, it is believed that there are currently 6 GD patients, 5 PD patients, 30 FD patients and around twenty MPS patients altogether. Meanwhile, all six GD patients are treated with Imiglucerase and none of the rest patients are receiving ERTs. Our study only includes around 50.8% (31/61) of the total samples. However, we believe the generalizability of our results is not limited due to the homogeneity of the patients with same ultra-rare disease. Another limitation is caused by recall bias and preference bias. Some patients could not remember the exact amount of their incomes as well as their expenditures. Also, some patients may overestimate their expenditures and underestimate their income to draw more attention from the local authorities and society. However, due to there is no available data of patients with the 4 LSDs from patient registry, claim database and health information system right now, our approach seems to be the most feasible way.

Conclusion

Based on the current policies in Shanghai and our study on the patients with four LSDs, few patients with LSDs in Shanghai could have access to available ERTs without a high reimbursement level. Though the cost-sharing mechanism of basic medical insurance, charity fund and patients had been explored for Gaucher disease in Shanghai, the OOP part still laid a heavy economic burden on the patients and their families. The healthcare security system should pay more attention to LSDs patients, which need to be treated with extremely expensive ERTs. The scope of drug reimbursement list and the reimbursement level should be further expanded and raised to help improve the quality of life of patients with LSDs.

Abbreviations

LSF: Lysosomal storage disease; ERT: Enzyme replacement treatment; GD: Gaucher disease; FD: Fabry disease; PD: Pompe disease; HST: Highly specialized technology; MPS: Mucopolysaccharidosis; NP: Niemann-Pick disease; NRDL: National Rare Diseases List; NDRL: National Drug Reimbursement List; QoL: Quality of life.

Declarations

Ethics approval and consent to participate

The survey was approved by the Medical Ethics Committee of Shanghai Health Development Research Centre (No. 2020004).

Consent for publication

Not applicable

Availability of data and materials

The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

Competing interests

The authors declare that they have no competing interests

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Authors' contributions

QK and CJ designed the survey questionnaire. DL, JH and HZ managed and coordinated the patient survey. JH and QK was in charge of collecting survey data and quality control. JH and LZ analyzed and interpreted the survey data. JH, LZ and QK contributed to write the manuscript. All authors read and approved the final manuscript.

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