LSD Brazil Network: Advancing the Diagnosis of Lysosomal Storage Diseases in Brazil and Latin America

Franciele Barbosa Trapp[1]; Mariluce Riegel[1] ; Marion Deon[1]; Franciele Lopes[1]; Gabrielle Iop[1]; Jessica Lamberty Faverzani[1]; Henrique Bordin Lucena Borges[1]; Marina Hentschke Lopes[1]; Inamara Moares[1]; Natasha Malvarezi[1]; Kristiane Michelin-Tirelli[2]; Fernanda Medeiros Sebastiao[2]; Osmar Rachor Toledo dos Santos[1]; Fernanda Rosa Pereira[1]; Roberto Giugliani[1]

1- Casa dos Raros, Porto Alegre, Brazil; 2- Medical Genetics Service, HCPA, Porto Alegre, Brazil;

BACKGROUND

The LSD Brazil Network (LBN) began in 2013, with the aim of making largely available tools to enable diagnosis of lysosomal storage diseases (LSDs), especially in Brazilian regions with poor access to diagnostic facilities. The LBN headquarter is in Porto Alegre, and can be accessed via website, email, toll-free telephone, or WhatsApp. The LSD network raises unrestricted grants from several sources to maintain its infrastructure and provide the diagnostic service free of charge. Now, after 12 years of LBN, the data obtained from 2013 to 2024, as well as overall description of LBN (structure and services) are presented in this work.

MATERIAL AND METHODS

From 2013 to 2024, biological samples from 29,942 patients were received and processed by laboratories affiliated to LBN, coming from all Brazilian regions, from some other countries, mainly Latin American. Figure 1 illustrates the mode of operation of the LSD Brazil Network.

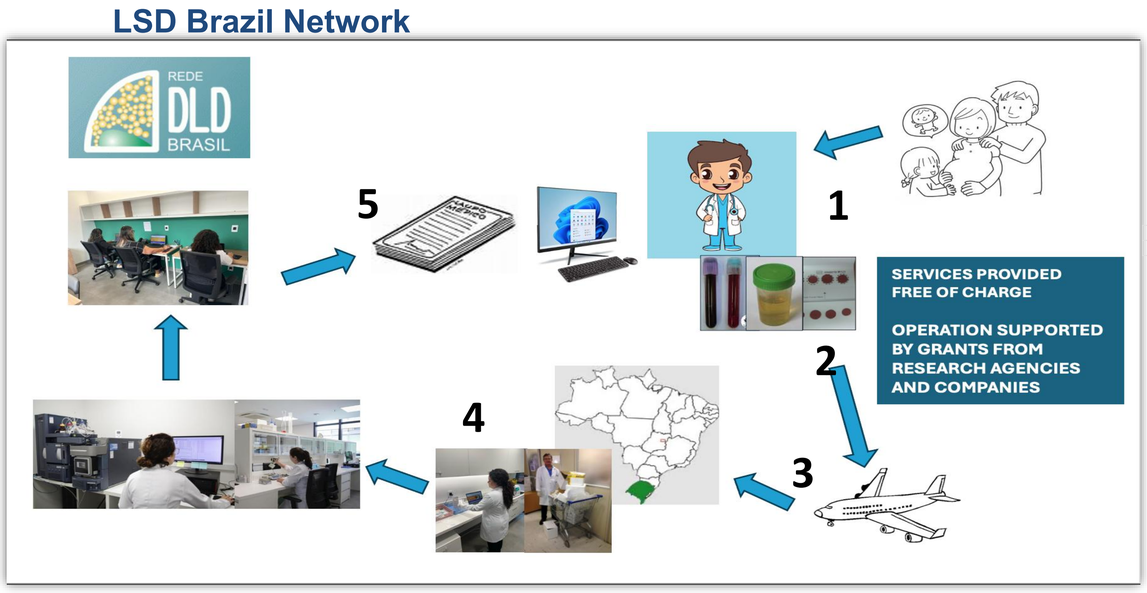

Figure 1. Schematic representation of the operation mode of LSD Brazil Network. 1-3. The requester (medical geneticist, metabolic pediatrician, or another specialist) who suspect a lysosomal disease in a patient, fills a form with basic clinical information and send it to the LBN by courier, along with an Informed Consent Form, and the appropriate samples (DBS, plasma, urine, CSF, etc.). 4.The diagnostic investigation is performed in several laboratories affiliated to the program, all located in Porto Alegre, Brazil. 5.When the investigation is completed, the requesting doctor can access a detailed report in the restricted area of the website.

RESULTS

Over the last 12 years, a total of 29,942 patients from Brazil and from some other countries (mainly Latin American) were investigated by LBN for an extended group of 27 LSDs and a total of 1,516 patients were detected (yield of 5%).

Table 1. Number of diagnoses and number of requests, and diagnosis rate per year.

Period	Number of diagnoses	Number of requests	Rate(%)
2013	146	2470	5.91
2014	129	2660	4.84
2015	113	3415	3.31
2016	156	3911	3.98
2017	142	3779	3.75
2018	150	2620	5.72
2019	143	2487	5.74
2020	100	1680	5.95
2021	142	1805	7.86
2022	99	1662	5.95
2023	102	1833	5.56
2024	94	1620	5.80
Total	1516	29.942	100

Table 2. Type and number of LSD diagnosed from 2013 to 2024.

Lysosomal Storage Diseases	Number of cases
Acid Sphingomyelinase Deficiency	83
Alpha-Mannosidosis	17
Fabry disease	34
Galactosialidosis	4
Gaucher disease	191
GM1 Gangliosidosis	19
GM2 Gangliosidosis - Sandhoff	12
GM2 Gangliosidosis - Tay-Sachs including B1	78
Krabbe Disease	76
Lysosomal Acid Lipase Deficiency	16
Metachromatic Leucodystrophy	51
Mucopolysaccharidosis type I	99
Mucopolysaccharidosis type II	187
Mucopolysaccharidosis type IIIA	37
Mucopolysaccharidosis type IIIB	60
Mucopolysaccharidosis type IIIC	42
Mucopolysaccharidosis type IIID	2
Mucopolysaccharidosis type IVA	136
Mucopolysaccharidosis type IVB	3
Mucopolysaccharidosis type VI	134
Mucopolysaccharidosis type VII	13
Multiple Sulfatase Deficiency	8
Neuronal Ceroid Lipofuscinosis type 1	11
Neuronal Ceroid Lipofuscinosis type 2	104
Niemann-Pick disease type C	37
Pompe disease	59
Sialidosis	3
TOTAL	1516

DISCUSSION AND CONCLUSIONS

From 2013 to 2024, a total of 1,516 cases of LSDs were diagnosed and the five most frequent diseases were), Gaucher (191), MPS II (187), MPS IVA (136), MPS VI (134), CLN2 (104), (Table 2). This distribution may not reflect the actual relative prevalence of LSDs, as testing for some diseases are more frequently requested due to the availability of therapies (examples: Gaucher, MPS, CLN2). Also, some diseases (as Fabry) have several other diagnostic services available in the region, and so the cases identified by LBN may represent only a fraction of the total diagnoses.

In summary, the LBN demonstrated to be a useful tool to enable the diagnosis of LSDs, especially in a region with scarce facilities as Brazil and Latin America.

CONFLICTS OF INTEREST. The authors declare no conflicts of interest.

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https://cdr.org.br/
contato@cdr.org.br
+55 (51) 3455-0000
Rua São Manoel, 730, 90620-110
Porto Alegre, Rio Grande do Sul, Brazil