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Research Article

Gaucher Disease: Clinical, Biochemical and Molecular Profile from Tertiary Care Center, Rajasthan

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ABSTRACT

Introduction: Gaucher disease (GD) is the most common inherited lysosomal storage disease, inherited as autosomal recessive and caused by mutations in acid ß- glucosidase *(GBA)*gene resulting in accumulation of ß- glucocerebroside in the macrophagic cells of the reticuloendothelial system.

Aims and objectives: To study clinical spectrum, biochemical and molecular profile in a cohort of GD patients in Rajasthan.

Material and methods: This hospital based observational study was carried out in department of pediatric medicine, SPMCHI, SMS Medical College, Jaipur from May 2020 to Aug 2021. Demographic and clinical data, investigations, ß- glucocerebrosidase enzyme activity, pathogenic mutation in GBA gene and anthropometry and biomarkers for monitoring treatment were collected.

Results: Thirty patients aged between 1 day and 18 years were studied. Out of 30 patients 26 patients were diagnosed with Type 1(non-neuronopathic) and 4 with Type 3(chronic neuronopathic) GD. Common presentation was hepatosplenomegaly, bicytopenia (anemia and thrombocytopenia) and growth failure. Bone disease in the form of bone pain in seven patient and osteomyelitis in one patient was identified. The most common GBA variant was c.1448T>C(p.Leu483pro) which was detected in 25 patients. ERT was received by 2 patients through charitable access program on compassionate basis. Hematologic and visceral manifestations were reversed and quality of life was improved following ERT.

Conclusions: Early identification and treatment improves quality of life in these patients therefore developing countries like India need to channelize funding and resources for definitive management and all health care providers should be made aware of the presenting signs and symptoms of GD.

Keywords: Gaucher Disease, GBA Gene Mutation, Hepatosplenomegaly, Enzyme Replacement Therapy (ERT).

INTRODUCTION

Gaucher disease (GD) is a hereditary autosomal recessive disease due to congenital deficiency of lysosomal enzyme acid β- glucosidase resulting in accumulation of nondegraded glycosphingolipids (glucocerebroside) in the macrophagic cells of the reticuloendothelial system.[1] It is caused by mutation in *GBA* gene, located on chromosome 1q21.[2]

Incidence of gaucher disease in general population is 1:40000 to 1:60000 and varies from 1 in 450 to 1 in 1000 in Ashkenazi Jews. [1] GD is characterised by anemia, thrombocytopenia, hepatosplenomegaly, growth failure, seizures and bone disease .[3]

GD is classified into type 1 (non-neuronopathic), type 2 (acute neuronopathic) and type 3 (subacute or chronic neuronopathic) based on age of onset and presence or absence of neurological symptoms.[4] The most prevalent is the type 1 which is characterized by anemia, thrombocytopenia, enlargement of liver and spleen, skeletal abnormalities (diffuse bone pain, osteolytic lesions, pathological fractures, avascular necrosis with subsequent joint collapse affecting proximal and distal femur, proximal tibia and proximal humerus, Erlenmeyer flask deformity) [5,6,7]. Some patients with type 1 GD also have pulmonary involvement with interstitial lung disease and pulmonary hypertension[8,9]. Type 2 GD is an acute neuronopathic form characterized by neurological involvement along with visceral symptoms. It is a severe form with onset in infancy and survival limited to first two or three years of life[10]. The neurological manifestations

involve oculomotor abnormalities (strabismus, saccade initiation abnormalities), bulbar palsy, seizures, hypertonia, swallowing difficulty and brain stem involvement[11,12,13]. Type 3 GD is also characterized by neurological involvement but symptoms appear later in life and include oculomotor apraxia, seizures, ataxia and dementia, with patients surviving until their third or fourth decade.[14]

GD is progressive disease and if not treated may lead to severe morbidity due to haemorrhage and skeletal complications, liver failure, pulmonary hypertension and sepsis, causing a reduction in quality of life and life expectation.[4]

Diagnosis of GD is based on clinical manifestations, physical features, laboratory test (blood counts, liver function tests, biomarkers- chitotriosidase) and confirmed by determination of acid β glucosidase enzyme activity in peripheral leucocytes and genetic mutation studies. [15]Mutation analysis helps in confirmation of diagnosis and is necessary for prenatal diagnosis in subsequent pregnancies for the families with an affected child.

Current treatments for GD, including enzyme replacement therapy (ERT) and substrate reduction therapy (SRT), can reverse many of the non-neurological manifestations including hepatosplenomegaly, cytopenia, bone pain and bone crisis, especially if administered in early stages of disease.[16]

The rationale of the study: Limited epidemiological data are available on Gaucher disease in India. Our study illustrates the clinical profile, genotype and management of Gaucher disease in a cohort of GD patients in Rajasthan. This could help in early identification of patients with Gaucher disease and timely initiation with management.

MATERIALS AND METHODS

Study Design and Setting

This was a hospital-based observational study conducted in the Department of Pediatrics, SPMCHI, SMS Medical College, Jaipur, Rajasthan, India. The study period was from May 2020 to August 2021.

Study Population

Children aged up to 18 years with a confirmed diagnosis of Gaucher disease were included in the study.

Inclusion Criteria

- Age up to 18 years
- Confirmed diagnosis of Gaucher disease based on:
    - Reduced beta-glucocerebrosidase enzyme activity on dried blood spot (DBS)
    - Identification of pathogenic mutation in the GBA gene

Exclusion Criteria

- Age above 18 years
- Refusal to give informed consent

Sample Size

Sample size was calculated using a 95% confidence interval and an alpha error of 0.05. Considering an expected 90 percent prevalence of splenomegaly in Gaucher disease and an allowable error of 11 percent, the required sample size was estimated to be 30 patients.

Data Collection and Clinical Evaluation

After obtaining written informed consent, detailed demographic, clinical, and family history was recorded. Clinical evaluation included presenting complaints such as fatigue, bleeding, recurrent infections, abdominal distension, bone pain, delayed puberty, seizures, vomiting, diarrhea, and any other relevant symptoms. Past history, developmental history, immunization status, and family history including consanguinity were documented.

Physical examination included vital signs, anthropometry (height, weight, head circumference), general physical examination, and systemic examination with emphasis on abdominal and neurological evaluation.

Investigations

Baseline investigations performed at diagnosis included:

Hematological and biochemical tests:

- Hemoglobin
- Total leukocyte count
- Platelet count
- Liver function tests (SGOT, SGPT, bilirubin, total protein, albumin)
- Serum calcium, phosphorus, alkaline phosphatase
- Serum LDH

Disease-specific investigations:

- Plasma chitotriosidase level
- Beta-glucocerebrosidase enzyme activity on dried blood spot (DBS)
- GBA gene mutation analysis

Imaging studies:

- Ultrasonography of abdomen for liver and spleen size
- Skeletal imaging or radiographs when indicated

Treatment and Follow-Up

Information regarding treatment was recorded, including enzyme replacement therapy,

symptomatic management, or history of splenectomy. Patients who were receiving enzyme replacement therapy were followed up for six months. Follow-up assessment included evaluation of weight, height, hemoglobin level, platelet count, and liver and spleen size based on clinical examination and ultrasonography.

Statistical Analysis

Data were entered into Microsoft Excel and analyzed using appropriate statistical methods. Quantitative variables were expressed as mean and standard deviation or median and range. Qualitative variables were expressed as frequencies and percentages. Chi-square test was applied for categorical variables. A p-value less than 0.05 was considered statistically significant.

RESULTS

In our cohort of 30 patients with gaucher disease, twenty-four were males and six were females. Twenty-six patients were identified as Type 1(non-neuronopathic) gaucher disease and four with type 3 (chronic neuronopathic) gaucher disease. No cases were having type 2 gaucher disease **(Fig 1)**.

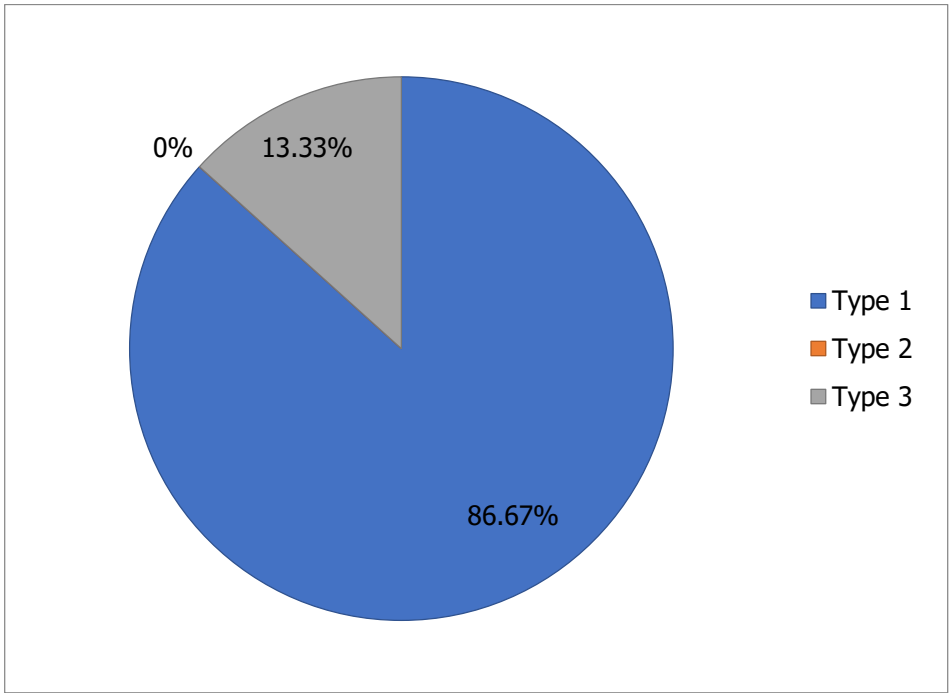

Figure 1. Distribution of Study Subjects According To Type of Gaucher Disease

In our study group mean age of presentation was 4.03 +_4.19years with a median of 2**(table 1)**. 21(70%) cases out of 30 patients had consanguinity and 5(16.6%) had a family history of gaucher disease in sibling.

Table 1. Distribution of Study Subjects According To Age at Presentation

Age at presentation	N	Percentage
≤1 years	11	37
1 – 5 years	9	30
6 – 10 years	7	23.3
> 10 years	3	10
Total	30	100
Mean ± SD	4.03 ± 4.19 years	
Median (Range)	2 (0.58 – 15 years)	

Most common clinical manifestation was abdominal distension (28/30) followed by mucosal bleeding (10/30), frequent infection (10/30), bone pain (7/30), fever (6/30), vomiting (4/30), delayed puberty (1/30), seizures (1/30), constipation (1/30) and diarrhoea (1/30) **(fig-2)**. Out of 30 patients, 20 (67%) patients had short stature and 17(56%) patients were undernourished.

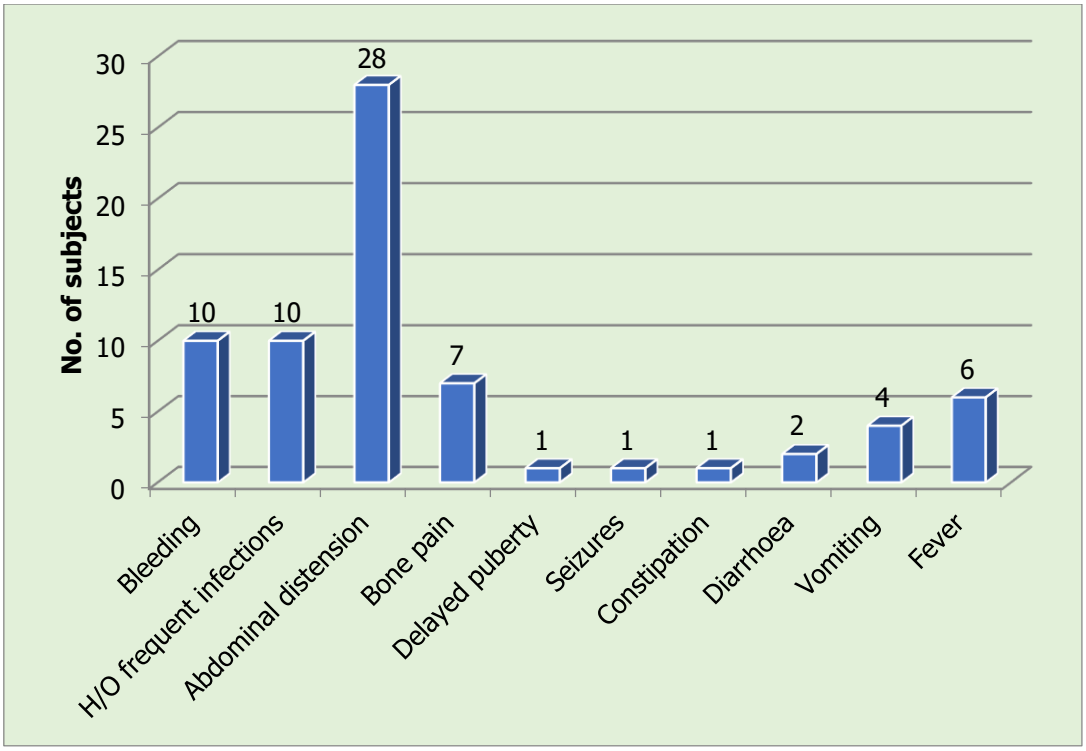

Figure 2. Presenting Complaints among Study Subjects

The most common clinical finding among type 1 GD group was splenomegaly (100%) followed by hepatomegaly (96.1%). Skeletal involvement was seen in 7(26.9%) patients.

Among 4 patients with type 3 GD, splenohepatomegaly was found in all patients and neurological involvement included epilepsy in 1/4 (25%), squint in 1/4(25%), oculomotor apraxia in 1/4 (25%) and cognitive deficits in 2/4(50%).

Haemoglobin and platelets were markedly reduced with a mean of 6.80±1.93 (range, 3.5 to 10.4g/dl) and 0.79±0.39 (range, 0.12 to1.5lakhs/dl) respectively at the time of presentation. Serum aspartate and alanine transaminases levels were 62.23±29.75 and 43.35±37.32 respectively. Plasma chitotriosidase levels ranged from 228.82 to 59922.01 with a median of 999.95 and a mean of 3757.76±10961.67nmol/ml/hr. GBA activity on DBS among 30 cases ranged from 0.46 to 1.9nmol/hr/mg protein with a median of 0.94 and a mean of 0.913±0.436nmol/hr/mg protein. Laboratory investigations at the time of presentation are depicted in **Table 2**.

Table 2. Blood Parameters among Study Subjects

	Mean ± SD	**Median (range)**
Haemoglobin (g/dl)	6.80 ± 1.93	7.25 (3.5 – 10.4)
TLC (x10^3/dl)	4.84± 2.12	4.4 (2.2 – 12.29)
Platelet (lakh/dl)	0.79 ± 0.39	0.79 (0.12 – 1.5)
SGOT (IU/L)	62.23 ± 29.75	50 (24 – 150)
SGPT (IU/L)	43.35 ± 37.32	26.5 (12 - 146)
SLDH	297.3 ± 88.57	290 (188 – 580)
Total bilirubin (mg/dl)	0.96 ± 0.40	0.86 (0.46 – 2.6)
Total protein (g/dl)	5.92 ± 0.32	5.9 (5.2 – 6.8)
Albumin (g/dl)	3.49 ± 0.29	3.5 (2.8 – 3.9)
Calcium(mg/dl)	8.23 ± 0.68	8.4 (6.8 – 9.5)
Phosphorus(mg/dl)	3.85 ± 0.54	3.95 (2.7 – 4.8)
ALP	259 ± 106.2	220 (108 – 490)
Chitotriosidase(nmol/ml/hr)	3757.76 ± 10961.67	999.95 (228.82 - 59922.01)
GBA activity (nmol/hr/mg protein)	0.913 ± 0.436	0.94 (0.46 – 1.9)

Out of 26 patients with non-neuronopathic type 1 GD, most common genetic mutation was homozygous L444P (88.4%). Compound heterozygous mutations were also found. Among four patients with chronic neuronopathic type 3 GD, homozygous L444P mutation was found in 2 patients followed by compound heterozygous mutation of (L444P)+(A456P) and (L444P)+(F2131) in the other two patients **(Table 3).**

Table 3. Molecular profile of Gaucher disease patients (n=30)

GBA mutation	Type 1		Type 3		Total	
	N	%	N	%	N	%
(L444P)+(L444P)	23	88.4	2	50	25	83.33
(L444P)+(A456P)	1	3.8	1	25	2	6.67
(L444P)+(F2131)	0	0.0	1	25	1	3.33
(L444P)=(IVS2+1G>C)	1	3.8	0	0	1	3.33
(R496C)+(R496C)	1	3.8	0	0	1	3.33
Total	26	100	4	100	30	100

In our cohort of 30 patients, 2 (6.67%) received enzyme replacement therapy (ERT), 10(33.33%) were given symptomatic and supportive treatment and 14(46.67%) did not receive any treatment due to lost to follow up. Four patients (13.33%) had undergone splenectomy from various centres **(Fig.3).**

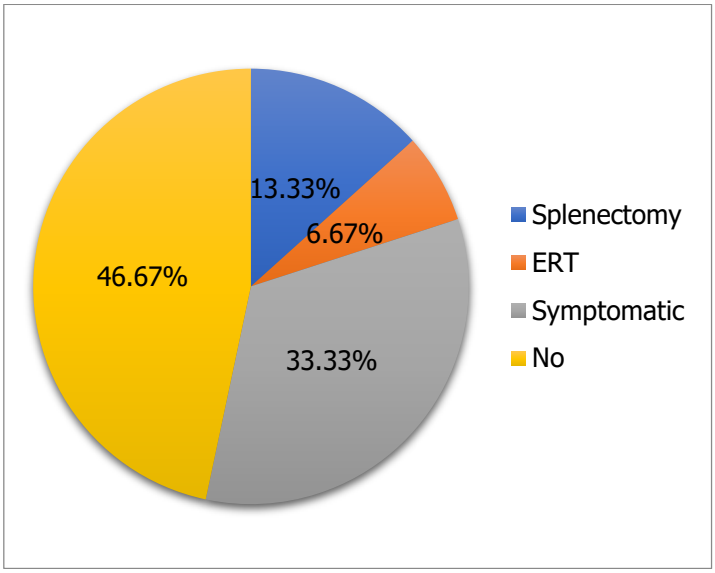

Figure 3.Treatment Given To Patients with Gaucher Disease

Follow up of patients on Enzyme Replacement Therapy

In our cohort of 30 patients, two were on ERT through charitable access program on compassionate basis. After ERT improvement in physical activity, weight, height, haemoglobin and platelet count was noted in both patients. Liver and spleen also regressed in size in both patients. **(Table 4)**.

Table 4. Change in Patient Characteristic after ERT

		Patient 1	Patient 2
Weight	Baseline	10 Kg	21 Kg
	After ERT	13 kg	25 Kg
Height	Baseline	88 cm	116 cm
	After ERT	90.5 cm	118 cm
Haemoglobin	Baseline	9.9 g/dl	10.4 g/dl
	After ERT	11.8 g/dl	11.6 g/dl
Platelet count	Baseline	1.16 lakh/dl	0.76 lakh/dl
	After ERT	1.56 lakh/dl	1.70 lakh/dl

Liver size	Baseline	8 cm	9 cm
	After ERT	6.5 cm	7 cm
Spleen size	Baseline	7 cm	9 cm
	After ERT	5 cm	7.5 cm

DISCUSSION

Gaucher disease is the most common lysosomal storage disorder (LSD) due to biallelic pathogenic variant in *GBA* gene causing deficiency of lysosomal enzyme acid β-glucosidase.[17] The human *GBA* gene, located on chromosome 1q22, has a highly homologous pseudogene sequence located 16 kb downstream. These include missense and nonsense mutations, splice junction mutations and complex alleles carrying two or more mutations, small insertions or deletions resulting in frameshifts or in-frame alterations.[18,19]

Among the three phenotypic variants of GD, type 1 is the most common, and reported predominately in Ashkenazi Jewish population. Type 2 is pan ethnic, while type 3 is limited to the Norrbottnian population in Sweden.[20,21]

A few Indian studies are available regarding screening, diagnosis and management of Gaucher disease in Indian population. The diagnosis of GD still remains challenging due to extreme variability of clinical manifestations, limited diagnostic facilities and lack of awareness among clinicians in resource limiting setting such as India. In our study we included 30 GD patients and studied their clinical manifestations, genetic characteristics and various treatment modalities they received at a tertiary care centre in Rajasthan.

In our study out of 30 cases, 26(86.67%) cases were of Type 1 and 4(13.33%) of type 3 gaucher disease. The mean age of presentation was 4.03±4.19 years and mean age of diagnosis was 5.98 **±** 4.38 years out of which 80% were males and 20% were females. Findings were similar to a study conducted by Himangi et al. in which the mean age at presentation was 4.6 ± 0.475 years and the mean age of diagnosis was 4.8 ± 0.188 years[22]. In a study conducted by Muranjan et al.[23] 45.4% GD patients had type 1 disease and 41% had type 3 GD with mean age of presentation of 56 (3.5-228) months. Barney AM et al. [17]conducted a study in 60 subjects with gaucher disease in India out of which males were 51.7% and females were 48.3% and median age of presentation was 1(0.7-5.7) year and 80% were of non neuronopathic type 1, 17% of type 3 and 3% of Type 2 GD.

In our study type 2 patients were not diagnosed may be because of early death in these patients before reaching higher centre or because considering gaucher disease as a differential diagnosis for neurological symptoms as a remote possibility.

The most predominant clinical manifestation in our study was abdominal distension in 93.3% (due to splenohepatomegaly) followed by fatigue, tiredness, bleeding and history in frequent infections in 33% (due to pancytopenia). Skeletal abnormalities were seen in 7/30, epilepsy in 1/30, squint in 1/30, oculomotor apraxia in 1/30 and cognitive deficits in 2/30. These findings were similar to that of Lee J Y et al.[24] and ICGG registry[25]. Barney AM et al.[17] conducted a study on 60 patients of gaucher disease out of which 6(10%) had hypotonia, 6(10%) had respiratory involvement,5(8.3%) had bone pain,20(33.3%) had developmental delay, 4(6.6%) cases had gaze palsy/saccades,9(15%) had seizures, 6(10%) had ecchymosis and 6(10%) had epistaxis and all had splenomegaly and hepatomegaly. In our study neurological manifestations were less and visceral and haematological manifestations were more because most of the patients belonged to non-neuronopathic gaucher disease.

Plasma chitotriosidase levels ranged from 228.82 to 59922.01 with a median of 999.95 and a mean of 3757.76±10961.67nmol/ml/hr. GBA activity on DBS among 30 cases ranged from 0.46 to 1.9nmol/hr/mg protein with a median of 0.94 and a mean of 0.913±0.436nmol/hr/mg protein. Similar findings with raised chitotriosidase levels and significantly reduced GBA activity were seen in studies by Sheth et al.[19] and Barney et al[17]. Increased chitotriosidase levels are also seen in LSD like Niemann- pick, cystinosis, fabry, GM1, krabbe, infectious diseases like malaria, tuberculosis, diabetes, sarcoidosis, β-thalassemia.

Lee JY et al[24] conducted study in 20 patients with gaucher disease in Korea in which haemoglobin level was 12.1±1.7mg/dl (range 9.4 to 14.6 mg/dl), platelet count was 72.25±30.06*10^3 /mm^3 (range, 16 to 315*10^3 /mm^3), SGOT levels were 53.9±50.0IU/L (range,11to225IU/L) and SGPT levels were 25.9±16.7IU/L (range 7 to 58IU/L). Plasma

chitotriosidase activity was markedly increased in 13 of 17 patients (76.5%) with mean of 3331.8± 2486.6nmol/hr/ml (range, 9.4 to 15918.1nmol/hr/ml). GBA activity decreased at 5.7±3.2pmol/min/mg (range, 0.2 to 11.6pmol/min/mg).

In our cohort the most common genetic mutation in Type 1 GD was found to be homozygous mutation of L444P (88.4%) followed by compound heterozygous mutation of L444P. Out of 4 patients of type 3 GD, 2 patients had homozygous mutation of L444P and other 2 had compound heterozygous mutation of L444P. Studies by Sheth et al.2019[19] and Muranjan et al.2016[23] have revealed the same.

In our study out of 30 patients, 2(6.67%) patients received ERT, 4(13.3%) had splenectomy, 10(33.33%) received symptomatic treatment and 14(46.67%) were lost to follow up. Of 2 patients who received ERT were followed up at 6 months as per consensus guidelines given by Ratna et al[26].Those receiving ERT showed improvement in well-being, weight and height, increase haemoglobin and platelet and reduction in liver and spleen size. ERT is costly drug and require fortnightly infusion for lifelong. ERT was received by 2 patients in our study (child of ESI employee, charitable access program). Our findings are consistent with study conducted in India by Barney AM et al[17] and Khalifa et al[27]. In resource limited settings such as in India, ERT is financially draining and not a sustainable option, despite being the standard of care treatment.

CONCLUSION

A high index of suspicion is required to diagnose rare genetic disorders like Gaucher Disease. Early identification and treatment improves quality of life in these patients therefore developing countries like India need to channelize funding and resources for definitive management and all health care providers should be made aware of the presenting signs and symptoms of GD.

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