

**Supplementary Table 1.** Comparison of clinical features which are highly predictive of Gaucher disease between patients with Gaucher disease and non-Gaucher disease.

Parameters	Gaucher disease (N=1)	Non-Gaucher disease (N=351)
Size of splenomegaly (cm), N; mean (SD)	1; 18.40	222; 14.56±3.39 SD
Presence of hepatomegaly	0	52 (14.81)
Specific abnormality in bone marrow biopsy	1; 1 (100.00)	160; 72 (45.00)
Signs and symptoms		
Splenomegaly	1 (100.00)	351 (100.00)
Thrombocytopenia	1 (100.00)	155 (44.16)
Anaemia	0	144 (41.03)
Hepatomegaly	0	52 (14.81)
Bone pain	0	14 (3.99)
Pathologic fracture	0	1 (0.28)
Osteonecrosis	0	1 (0.28)
Others	0	41 (11.68)
Duration of signs and symptoms (days), N; mean (SD)	1; 9842.00	1195.32±1959.65 SD
Family history of Gaucher disease	0	1 (0.28)
Laboratory data, N; mean (SD)		
Haemoglobin	1; 11.00	347; 12.22±2.73 SD
WBC	1; 4.12	348; 8.21±21.91 SD
RBC	1; 3.55	348; 4.07±0.92 SD
Platelet	1; 82.00	348; 164.11±143.25 SD
Serum iron	1; 58.50	102; 86.16±62.49 SD
Serum ferritin	1; 1673.90	135; 533.23±1098.82 SD
Transferrin	0	12; 144.94±123.09 SD
BUN	1; 13.00	295; 15.55±8.71 SD
Creatinine	1; 0.53	328; 0.97±1.15 SD
SGPT	1; 15.00 1; 36.00	332; 28.36±31.52 SD
SGOT	1; 1.55	332; 31.15±43.39 SD
Total bilirubin	1; 8.20	330; 1.10±1.20 SD
Total protein	1; 3.50	333; 6.95±0.83 SD
Albumin		333; 4.11±0.67 SD
Serum protein electrophoresis	0	N=205
Normal	0	20±9.76 SD
Hypogammaglobulinemia	0	2±0.98 SD
Monoclonal peak	0	5±2.44 SD
Other	0	5±2.44 SD
Not done	0	173±84.39 SD
Acid β-Glucosidase		
DBS Result		
Normal	0	331 (94.30)
Abnormal	1 (100.00)	15 (4.27)
Indeterminate	0	5 (1.42)
Activity value (nmol/hr/mg protein), N; mean (SD)	0.20	8.08±4.87 SD
Diagnosis		
Normal	0	306 (87.18)
Abnormal (Gaucher doubt)	1 (100.00)	5 (1.42)
Other (heterozygous, etc.)	0	40 (11.40)
Acid sphingomyelinase deficiency		
DBS Result		
Normal	0	188
Abnormal	0	186 (98.94)
Indeterminate	0	2(1.06)
Presence of mutation	1	7
Positive	1 (100.00)	0
Negative	0	7 (100.00)
Carrier	0	0

Data presented as N (%), unless otherwise specified.

Abbreviations: BUN, blood urea nitrogen; DBS, dry blood spot; RBC, red blood cells; SD, standard deviation; SGOT, serum glutamic-oxaloacetic transaminase; SGPT, serum glutamic-pyruvic transaminase; WBC, white blood cells.