Lysosomal Storage Disease in Iran (Report of Molecular Study)

How to Cite this Article: Houshmand M, Tonekaboni SH, Karimzadeh P, Aryani O, Ashrafi MR, Salehpour Sh, Badv Sh, Shakiba M, Alaee MR, Farshid Sh. Lysosomal Storage Disease in Iran. (Report of Molecular Study). Iran J Child Neurol Autumn 2012; 6:4 (suppl. 1): 22.

Massoud HOUSHMAND PhD[1], Seyed Hassan TONEKABONI, MD[2], Parvaneh KARIMZADEH MD[2], Omid ARYANi MD[3], Mahmoudreza ASHRAFI MD[4], Shadab SALEHPOUR MD[5], Shervin BADV MD[6], Marjan SHAKIBA MD[7], Mohammad Reza ALAEE MD[8], Shahla FARSHIDI MD[9]

1. Assistant Professor of Human Molecular Genetics, Department of Medical Genetic, National Institute for Genetic Engineering and Biotechnology, Tehran, Iran
2.Professor of Pediatric Neurology, Pediatric Neurology Research Center, Shahid Beheshti University of Medical sciences,Tehran, Iran
3. Genetic Counselor, Medical Genetic Dep. Special Medical Center, Tehran, Iran
4. Professor of Pediatric Neurology, Growth and Development Research Center, Children´s Medical Center, Tehran University of Medical Science, Tehran, Iran
5.Associate Professor of Pediatric Endocrinology and Metabolism, Shahid Beheshti University of Medical Sciences, Tehran, Iran
6.Assistant Professor of Pediatric Neurology, Zanjan University of Medical Sciences, Zanjan, Iran
7. Associate Professor of Pediatric Endocrinology and Metabolism, Shahid Beheshti University of Medical Sciences, Tehran, Iran
8.Associate Professor of Endocrinology, Shahid Beheshti University of Medical Sciences (SBMU), Tehran, Iran
9. Nasle Omid Hope Foundation

Corresponding Author:
Houshmand M. PhD
Medical Genetic Dep. Special Medical Center. Tehran, Iran
Email: Housh62@yahoo.com

Lysosomal storage disorders (LSDs) are relatively rare inborn errors of metabolism, resulting from the accumulation of substrates within the lysosomes. They represent a group of more than 40 distinct genetic disorders. Most of these disorders are inherited in an autosomal recessive manner, except Fabry's disease and mucopolysaccharidoses type II (MPS II) which are inherited in an X-linked recessive manner. Most disorders present clinically with multi-system involvement. Common clinical features involve bony dysplasia, hepatosplenomegaly, central nervous system dysfunction, haematological abnormalities, and coarse hair and facial features. There are many phenotypical similarities within the categories. Potential treatments for some of these disorders are available in the form of enzyme replacement therapy and bone marrow transplantation. PCR-sequencing methods were used for genetic investigation of 236 pediatric cases referred or diagnosed in our department over a period of 3 years from Nov 2009 to Nov 2012. Detailed clinical data, including sex, age of onset of disease, age at diagnosis, mode of presentation, family history, consanguinity rates, and high-risk screening results were collected. Biochemical analysis was done in different laboratories in abroad.

Keywords: Genetic; lysosomal storage disease; Child; Iran

Diagnosis	No	%
Gaucher disease	29	12.3
Niemann-Pick disease A and B	28	11.9
Metachromatic leukodystrophy	14	5.9
Krabbe disease	5	2.1
GM1 Gangliosidosis	12	5
Tay-Sachs disease	31	13
Sandhoff disease	12	5
Mucopolysaccharidoses	74	31.4
I-Cell disease/Mucolipidosis II	3	1.3
Lysosomal glycogen storage disease-Pompe's disease	5	2.1
Activator Deficiency/GM2 Gangliosidosis	2	0.8
Alpha-mannosidosis	2	0.8
Cystinosis	3	1.3
Fabry disease	1	0.4
Farber disease	2	0.8
Fucosidosis	2	0.8
Neuronal Ceroid Lipofuscinoses	8	3.4
Wolman disease	3	1.3
Total	236	100