Tehran University of Medical Sciences

Twenty-Eighth International Congress of Pediatrics
19 October to 21 October 2016
Tehran

Molecular analysis of *ARSB* gene in Iranian patients affected by Maroteaux–Lamy syndrome

Kouroshnia A. [1](MS), Davoudi-Dehaghani E. [1,2] (PhD), Abiri M. [3] (PhD), Dabbagh-Bagheri S. [1] (BS), Shirzad T. [1] (BS), Jamali M.[1] (MS), Bagherian H. [1] (MD), Zeinali S.[1,2](PhD)

[1] Kawsar Human Genetics Research Center, Tehran, Iran.
[2] Department of Molecular Medicine, Biotechnology Research Center, Pasteur Institute of Iran, Tehran, Iran.
[3] Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences, Tehran, Iran.

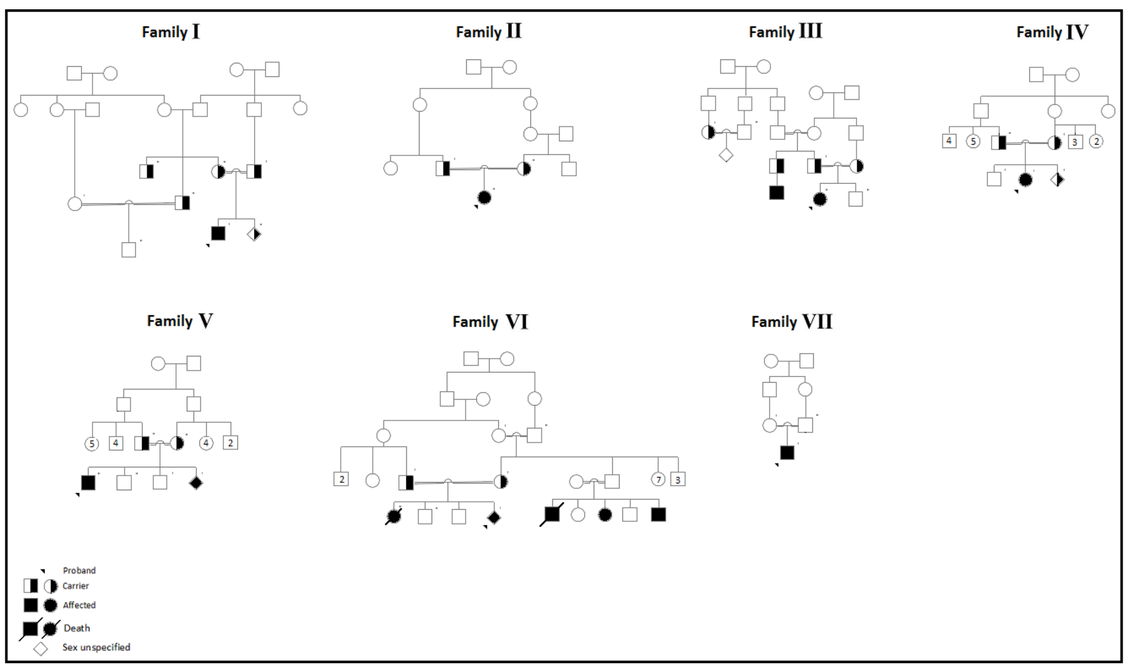
Responsible Writer: Zeinali S. (PhD), Department of Molecular Medicine, Biotechnology Research Center, Pasteur Institute of Iran, Tehran, Iran. Kawsar Human Genetics Research Center, Tehran, Iran.

Introduction:

Mucopolysaccharidosis VI (MPS VI) or Maroteaux–Lamy syndrome is an autosomal recessive lysosomal storage disorder. This abnormality is caused by mutations in *ARSB* gene. Affected individuals experience a lot of problems such as coarse facial features, cloudy corneas, skeletal abnormalities, growth retardation, enlarged organs, and deafness. Skeletal deformities as a main clinical symptom can be associated with joints and hip pains and also difficulty in walking. These problems bother patients by causing movement limitations.

Design/ Method:

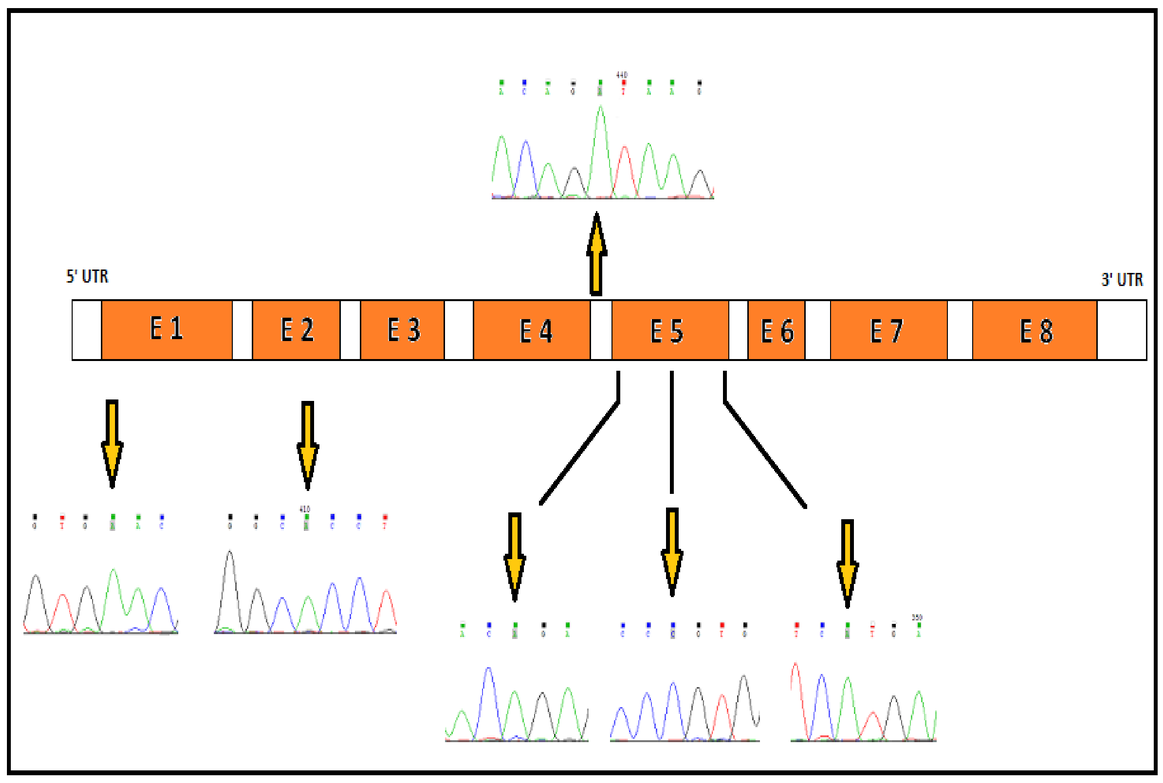
A total of 8 patients from 7 different families were included in this study. DNA extraction was performed using salting out method. Mutation detection was performed by Sanger sequencing of all coding exons and boundary regions of the *ARSB* gene. In silico studies were performed using PolyPhen-2, SIFT and mutation taster.



Pedigrees of the MPS VI families

Result :

Three novel and three previously reported mutations were identified in exons 1, 2, 4 and 5 of the *ARSB* gene. Five of six mutations were missense mutation and only one mutation was splicing mutation. In silico prediction and segregation analysis of mutations in families showed that the identified mutations can cause MPS VI in these families.



A schematic representation of 8 exons in the ARSB gene and identified variants in this study

Discussion:

The Maroteaux–Lamy syndrome shows genetic heterogeneity within Iranian population. However more studies in Iran can help to identify the most common mutations in this gene in this country.

References:

1. Mathew, Juby, Sujatha M. Jagadeesh, Meenakshi Bhat, S. Udhaya Kumar, Saravanamuthu Thiyagarajan, and Sudha Srinivasan. "Mutations in ARSB in MPS VI patients in India." Molecular genetics and metabolism reports 4 (2015): 53-61.

2. Valayannopoulos, Vassili, Helen Nicely, Paul Harmatz, and Sean Turbeville. "Mucopolysaccharidosis VI." Orphanet journal of rare diseases 5, no. 1 (2010): 1.