A Rare Case Of Mucopolysaccharidosis Type VI (Maroteaux–Lamy Syndrome)

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ICNC 2022
17th INTERNATIONAL CHILD NEUROLOGY CONGRESS
ANTALYA, TURKEY | OCTOBER 3-7, 2022

Abstract

Objectives: Mucopolysaccharidosis type VI or Maroteaux–Lamy syndrome is a rare, autosomal recessive lysosomal storage disorder caused by deficient enzymatic activity of N-acetyl galactosamine-4-sulphatase, which is caused by mutations in the arylsulphatase B gene. A case of a child with mucopolysaccharidosis type VI is reported. Methods: A 16/12-year-old Turkish girl presented with developmental delay. The female child was born to healthy, consanguineous parents as their third child. One of her siblings is healthy and the other is being followed up with a diagnosis of epilepsy. On examination she had short stature, short neck, coarse facial features, open mouth, macroglossia, a depressed nasal bridge and small stubby fingers, a low arched palate was observed. There was no evidence of hepatosplenomegaly. Psychomotor and speech development was mildly delayed. The parents noticed intermittent, third finger contractions which could be returned to normal posture with mother's help. Abdominal ultrasonography and skeletal survey revealed normal. Echocardiography showed 2 -3. degree mıtral valve insufficiency, mitral and aortic valve prolapsos. Ophthalmology assessment showed mild corneal clouding. Results: Based on clinical and laboratory findings mucopolysaccharidosis was suspected and confirmed by analysis of glycosaminoglycans in urine. The study of lysosomal enzymes showed a sharp decrease in the activity of Arilsülfataz B in the blood (<0,1 nmol/mg/h). Conclusion: It is possible to diagnose a case of mucopolysaccharidosis based on clinical findings and radiological features. Enzymatic studies with a careful and systemic approach are required for definitive type diagnosis.

Objectives

Mucopolysaccharidosis type VI also known as Maroteaux–Lamy syndrome, is a rare, autosomal recessive lysosomal storage disorder caused by deficient enzymatic activity of (arylsulfatase B), which leads to the accumulation of glycosaminoglycans (GAGs) throughout the body, particularly dermatan sulfate. Although MPS VI is a rare disease, with an estimated incidence of 1.3–4.5 in 100,000 births, it is characterized by a heterogeneous clinical, radiological and genetic.

Methods

A 16/12-year-old Turkish girl presented with developmental delay. The female child was born to healthy, consanguineous parents as their third child. One of her siblings is healthy and the other is being followed up with a diagnosis of epilepsy. On examination she had short stature, short neck, coarse facial features, open mouth, macroglossia, a depressed nasal bridge and small stubby fingers, a low arched palate was observed. There was no evidence of hepatosplenomegaly. Psychomotor and speech development was mildly delayed. The parents noticed intermittent, third finger contractions which could be returned to normal posture with mother's help.

Abdominal ultrasonography and skeletal survey revealed normal. Echocardiography showed 2 -3. degree mıtral valve insufficiency, mitral and aortic valve prolapsos. Ophthalmology assessment showed mild corneal clouding.

Results

Based on clinical and laboratory findings mucopolysaccharidosis was suspected and confirmed by analysis of glycosaminoglycans in urine. The study of lysosomal enzymes showed a sharp decrease in the activity of Arilsülfataz B in the blood (<0,1 nmol/mg/h).

Famıly permıssıon has been taken for photos

Conclusion

The age of presentation and clinical manifestations of MPS VI may greatly vary. The spectrum of clinical phenotypes may include a large head, coarse facial features, thick eyebrows, ocular proptosis, broad nasal bridge, thick lips, ogival palate, badly implanted teeth, macroglossia, short neck, narrow shoulders, globular abdomen, fingers and toes in a semi-fexion position, single palmar fold, short stature, dysostosis multiplex, generalized hypertrichosis, valgus knees, fat and thick feet, and less elastic skin. Despite the clear physical changes, the disease does not cause intellectual disability. Cardiac problems includes heart valve abnormalities, respiratory abnormalities, hepatosplenomegaly, umbilical-inguinal hernia, glaucoma and progressive corneal opacification, increased intracranial pressure, recurrent ear infections and hearing loss may occur.

As with most genetic conditions, there is no cure for MPS VI yet. Two therapeutic strategies have been used to reduce disease manifestations and improve patients' quality of life: HSCT and ERT. There is increasing evidence that early initiation of ERT in younger patients has clear benefits in preventing the progression of major disease symptoms. It is possible to early diagnose a case of mucopolysaccharidosis based on clinical findings and radiological features. Enzymatic studies with a careful and systemic approach are required for definitive type diagnosis

References

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