PRIDOSTIGMIN TREATMENT IN X-LINKED MYOTBULAR MYOPATHY

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INTRODUCTION

Myotubular myopathy (MTM) is a rare congenital neuromuscular disease characterized by neonatal hypotonia, muscle weakness and respiratory distress. The most common and severe type is X-linked myotubular myopathy (XLMTM) which is known to be caused by a mutation in the MTM1 gene at the Xq28 locus, which encodes the myotubular protein. Prenatal history includes polyhydramnios and decreased fetal movement. Facial weakness, ptosis, and ophthalmoplegia are also quite often. Individuals with X-linked myotubular myopathy usually dies in the early neonatal period due to respiratory failure. In this case, we discussed about genetically determined XLMTM and the clinical improvement it gave to pyridostigmine treatment of a patient whose etiology of hypotonia is being investigated.

CASE:

A male baby born to 30 yr old mother at 37-weeks of gestational age via cesarean section was apneic and after ventilation was performed with a bag valve mask for 1 minute spontaneous breathing started. First and 5th minute Apgar scores were 5 and 7, respectively. Umblical cord blood gas values were normal. When the antenatal history was deepened, fetal movements were decreased in the last trimester and polyhydramnios was present. There was no consanguinity between the parents. Deep tendon reflexes were absent, sucking reflex was weak, and swallowing dysfunction was present. There was spontaneous, equal but decreased motion in all four limbs. There were no signs of ptosis and ophthalmoplegia. Bilateral undescended testis was present.

Patient was intubated and could not be extubated at follow-up. Metabolic work up labs, SMA gene analysis, Cranial MR and MRS were normal. It was suspected to be neuromuscular disease and clinical exome sequencing was sent, and MTM1 mutation was detected as a result. Pridostigmine treatment was initiated and the patient was extubated during the follow-up, and his extremity movements and swallowing function increased. He was discharged after being followed up.

RESULTS

X-linked myotubular myopathy is a rare congenital muscle disease characterized by hypotonia, muscle weakness and respiratory distress. Genetic diagnosis is available and important because it makes prenatal diagnosis possible.

In some MTM mutations, pyridostigmine can provide clinical improvement in patients by affecting both muscle and muscle nerve junctions which indicates that the disease may involve the neuromuscular junction as well as the muscle.

CONCLUSION

As in this case; pyridostigmine; In patients with MTM1 mutation, it can be an effective treatment to increase extremity movement, decrease the need for breathing apparatus and increase swallowing function.

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