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A CASE OF NEURO-MOTOR FAILURE AND SEIZURES RELATED TO *TPP1* MUTATION: CASE REPORT AND MANAGEMENT

Dipanjana Datta[1,2] and Rajeev Agarwal

[1]ATGC Diagnostics Pvt Ltd, India
[2]CARE IVF, India

A non-consanguineous couple married for 18 years visits our infertility clinic with the following history. The couple conceived through medical assistance followed by an unremarkable pregnancy and vaginally gave birth to a baby girl at full term. Till the 1st year the child reached all milestones but then delay in growth and development started. She had delay in speech as well. At 2 years, seizures started, and progressive degeneration of locomotor functions in the extremities was observed. Gradually she lost her coordination skills and doctor suspected of muscular dystrophy. No motor testing or biochemical or genetic testing was done. The daughter expired at 8 yrs following a seizure. The couple by that time had conceived naturally again. The couple delivered a full term baby daughter at birth. The baby had delayed cry at birth. All milestones were achieved till 3, then, the child started having seizures. Became hypotonic and EEG reports were abnormal. No other tests were conducted. The child died at 5 yrs. The couple again conceived naturally and gave birth to a full term baby boy. Pregnancy was unremarkable. The boy child reached all reached milestones at 3 years but soon after that started losing muscle tone. EEG conducted was observed to be abnormal. Chromosomal studies had nothing remarkable. The prescription of the paediatric mentions presence of dysmorphic features. Progressive degeneration of locomotors functions in the extremities. The baby boy expired at 5 yrs. The couple had conceived again naturally has a baby girl, full term vaginally delivered. The baby girl is of 4 years age, normal and has no symptoms. The couple seeks assistance in having a healthy child. The neurologists suspected mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes. Genetic counselling was done and three generation pedigree was constructed. The couple was advised for genetic testing for mitochondrial and nuclear coded genes. It was found that the couple was heterozygous for a mutation c.1547_1548delTT in *TPP1* gene. This was classified as a pathogenic gene and reported for neuronal ceroid lipofuscinosis 2 and Spinocerebellar ataxia 7. Here we report this mutation in association with the above mentioned phenotypes. The c.1547_1548delTT variant is a deletion of 2 nucleotides, resulting in frameshift and premature termination. This variant has been reported in an individual with late infantile neuronal ceroid lipofuscinosis1 and it is also of a type expected to cause disease. The c.1547_1548delTT *TPP1* variant is classified as pathogenic. The couple was advised to go for PGT-M in the embryos generated through IVF. The couple followed the management guidelines and the healthy embryo was implanted.

dipanjanad@gmail.com