EuroSciCon

EuroScicon Joint event on

Genetics, Cell and Gene Therapy

August 20-21, 2018
Amsterdam, Netherlands

Biochem Mol biol 2018 Volume: 4
DOI: 10.21767/2471-8084-C3-015

CLINICAL MANIFESTATION OF A VARIANT OF *FBP1* AND ITS ROLE IN INBORN ERRORS OF METABOLISM

Dipanjana Datta[1,2] and Rajeev Agarwal

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

A non-consanguineous couple conceived naturally and had a baby in 2011. The baby eventually died due to hyperbilirubinemia and associated symptoms. The child was reported to have diarrhoea and continuous motion for 7 days before he expired. The couple again conceived in 2013 and the child was born with cleft palate. Corrective surgery was done but eventually they lost the baby at 8 months due to vomiting and diarrhea for 21 days. The couple came to our clinic in hope that we could help them to have a healthy child. Our team after analyzing the discharge reports of the babies and following genetic counselling concluded that the death of the babies might be due to reasons related to inborn error of metabolism. We did a panel testing for inborn error of metabolism and we found a likely pathogenic variant in *FBP1* in the couple. We present this pathogenic variant in association with the symptoms in this communication.

dipanjanad@gmail.com