Case Report

India's First Child using PGT-M, PGT-A and HLA Matching for Helping a Sibling having β-Thalassemia Major

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ABSTRACT

β-thalassemia is a common single-gene disorder in India, with hematopoietic stem cell transplantation (HSCT) being the only cure. HSCT with matched unrelated donor is less successful, whereas finding a human leukocyte antigen (HLA)-matched related donor is difficult. Preimplantation genetic testing for monogenic diseases (PGT-M) with HLA matching is a novel option to have a matched sibling for HSCT for couples having an affected child. We present the first such case report in India. A couple, both carriers of β-thalassemia and having an affected son, underwent PGT-M with HLA matching combined with preimplantation genetic testing for aneuploidies of embryos to have a β - thalassemia-free child. This resulted in birth of a 10/10 HLA-matched sibling.

KEYWORDS: *Human leukocyte antigen matching, hematopoietic stem cell transplantation, preimplantation genetic testing for aneuploidies, preimplantation genetic testing for monogenic diseases, β-thalassemia*