



PREIMPLANTATION GENETIC TEST FOR AUTOSOMAL RECESSIVE HYPERINSULINISM AND CANAVAN SYNDROME - A CASE REPORT

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INTRODUCTION

Preimplantation genetic test (PGT) can assess embryonic aneuploidies (PGT-A), monogenic diseases (PGT-M) and chromosomal structural abnormalities (PGT-SR).

CASE REPORT

Young second cousins were referred to PGT-M due to a previous baby with Familial hyperinsulinemic hypoglycemia 1 (OMIM 256450), both heterozygous for c.4412-13G>A variant in ABCC8 gene.

Previous spontaneous pregnancy with premature rupture of membranes and vaginal delivery at 33 weeks resulted in large for gestational age boy (3.890g) in cardiac arrest; he presented maintained hypoglycemia, edema, and recurrent infections, undergoing dialysis and tracheostomy; death at 5 months.

A gene probe for PGT-M was performed. After 2 cycles of In Vitro Fertilization, 5 embryos were biopsied. On the day of embryo biopsy, the couple was informed about the confirmation of Canavan syndrome (OMIM 271900) in a cousin. A more comprehensive carrier test was then performed and showed that both were heterozygous for c.914C>A mutation in ASPA gene. After adapting the PGT-M protocol for both pathologies, it was observed: one unaffected embryo and 4 hyperinsulinemia carriers; 2 affected embryos and 3 Canavan syndrome carriers; 3 carrier embryos were euploid. One embryo was transferred, resulting in biochemical pregnancy; in another cycle, one embryo was transferred, resulting in an ongoing pregnancy.

DISCUSSION

This case reinforces the importance of a detailed family history and the need for comprehensive carrier screening in consanguineous couples. When both are carriers for a recessive disease, couples should be advised of the possibility of performing PGT-M to ensure transfer of an unaffected embryo.