

# Preimplantation genetic testing and HLA-matching to select a healthy donor for sibling haematopoietic stem cell transplantation for a child with beta-thalassaemia major: A case report

Di Ling Leong, Jang Jih Chen, Michelle Sheng Rong Lim, Pak Seng Wong

Sunfert Intentional Fertility Centre, Kuala Lumpur, Malaysia

## ABSTRACT

**Introduction:** A child with Beta thalassaemia major may require regular blood transfusions and iron chelation treatment leading to significant morbidity and costs. We report a case of in-vitro fertilisation (IVF) with pre-implantation genetic testing (PGT-M) for human leucocyte antigen (HLA) tissue typing to selectively transfer an embryo to have a child who may provide umbilical cord blood for transplantation to the affected sibling. **Case Description:** The patient was a 36-year-old woman who carried a beta globin gene mutation at CD26 while her husband carried a beta globin gene mutation at CD35. They have a beta thalassaemia major child that inherited both mutations from the couple. Following a standard stimulation cycle, five blastocysts were biopsied and were tested with Infinium® Karyomapping Assay (Illumina). One blastocyst was aneuploid and affected. The remaining four blastocysts were euploid. Of these, one was confirmed to be unaffected, but it was not HLA-matched with the affected child. The remaining three were heterozygous (carrier) for either mutation at CD26 or CD35 and of these, only one blastocyst was eventually found to be HLA identical with the affected child. This HLA-matched blastocyst of grade 3AB (Modified Gardner's grading) was then vitrified-warmed, transferred and a gestational sac was observed. **Discussion:** This successful outcome suggests that high-resolution PGT-M with Karyomapping is a powerful screening tool in preventing unwanted genes from being passed down to their child with an added benefit of HLA-matching in a single biopsy to assist in the search of a HLA-compatible donor to the affected child.

# A prospective observational study to assess the efficacy of oral progestogen in first trimester threatened miscarriage

Mazrin Nur MA<sup>1</sup>, Zalina Nusee<sup>1</sup>, Faridah MY<sup>2</sup>, Nasuha Yaacob<sup>2</sup>

<sup>1</sup>Kulliyyah of Medicine, International Islamic University Malaysia, Kuantan, Pahang, Malaysia, <sup>2</sup>Department of Obstetrics and Gynaecology, Hospital Sultanah Nur Zahirah, Kuala Terengganu, Terengganu, Malaysia

## ABSTRACT

**Introduction:** Miscarriage occurs in 15-20% of all clinically recognized pregnancies. The aim of this study was to establish the efficacy of oral progestogen (intervention group) compared to conservative management, which was bed rest (control group) in subjects with the first trimester threatened miscarriage. The primary outcome of this study was the number of miscarriages by 20 weeks of gestation. **Methods:** This prospective observational study was conducted at gynaecology ward and Early Pregnancy Assessment Unit (EPAU) of Hospital Sultanah Nur Zahirah and the Department of Obstetrics and Gynaecology, Sultan Ahmad Shah Medical Centre IIUM for ten months, from June 2019 until March 2020. A total of 217 pregnant women who presented with threatened miscarriage of less or equal to 12 weeks of gestation were recruited. In the intervention group, the subjects were given 40 mg (4 tablets) of dydrogesterone orally at once, then 10 mg (1 tablet) in the interval of every 8 h until the symptoms abate, while the subjects in the control group were advised for bed rest as conservative management. **Results:** The baseline demographic data in both studied groups were similar and there was no significant ( $p > 0.05$ ) difference in age, BMI, parity, and time interval to pregnancy in both groups. There was an insignificantly smaller number of miscarriages in the intervention group (23, 46%) compared to the control group (27, 54%). **Conclusions:** In conclusion, oral progestogen therapy was an ineffective way to reduce the number of miscarriages in patients with the first trimester threatened miscarriage.