Wernicke's encephalopathy secondary to hyperemesis gravidarum: A case report

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ABSTRACT

Introduction: Wernicke's Encephalopathy is a reversible acute neurological disorder which is a rare but known complication of Hyperemesis gravidarum due to thiamine deficiency The non-alcoholic prevalence varies from 0.04% to 0.13%. **Case Description:** A 31-year-old, primigravida at 15 weeks of gestation presented with confusion and vomiting for the past three months. During admission, the patient was giving incoherent history and not orientated to time, place, and person. The patient was diagnosed with Wernicke encephalopathy as evidenced by confusion, ocular abnormalities, and dysmetria. The diagnosis was further supported by MRI scan, which shows fairly symmetrical T2W / FLAIR hyperintensities at bilateral dorsomedial thalami, tectal plate and periaqueductal area, likely due to toxic-metabolic causes. She received ICU care, aggressive thiamine administration, and electrolyte correction, and was discharged with oral thiamine. **Discussion:** Wernicke's encephalopathy is characterised by a triad of cerebellar sign, confusion, and ophthalmoparesis with nystagmus, giving a sensitivity of 23%. European Federation of Neurological Societies (EFNS) suggested that by taking into consideration dietary deficiencies in addition to the classical triad, patients who had at least two of the four features would have an increased sensitivity of 85%. EFNS also recommended that 200 mg thiamine should be given three times daily via intravenous route before the commencement of the diet. Complete remission occurred in only 29% of patients and permanent residual impairment is common. There is also an increased risk of miscarriage, preterm birth, and intrauterine growth restriction. Overall pregnancy loss rate including fetal loss and termination was 48%. If left untreated, Wernicke's encephalopathy could worsen to Korsakoff syndrome.

Combined pre-implantation genetic testing for aneuploidy (PGT-A) and pre-implantation genetic testing for monogenic disorder (PGT-M) analysis from a single embryo biopsy: A Malaysian case with de novo mutation in the CACNA1S gene

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ABSTRACT

Introduction: A couple was subjected to in-vitro fertilization with PGT-M treatment at our centre. The male partner was tested to carry a *de novo* CACNA1S mutation and was diagnosed with Hypokalemic Periodic Paralysis, an autosomal dominant disorder. The parents and the siblings of the male partner are not affected, and this couple has not had any children. Therefore, there is no genetic reference available. PGT-M testing without reference is very difficult and requires tedious and technically complex molecular procedures to perform the test. We used targeted amplification and next-generation sequencing (NGS) technology with specialized probes designed specific to the mutation site. **Methods:** Through using mutational site analysis, we were able to identify affected and unaffected embryos without the use of a reference. The results were further verified with SNP analysis. After screening with PGT-M, the samples from identified unaffected embryos were further subjected to PGT-A without the need for a re-biopsy. Results: A Euploid and unaffected embryo was transferred and a successful pregnancy was achieved, with a gestational sac and fetal heart activity detected during the ultrasound scanning at 5 weeks gestation. **Conclusion:** We successfully performed PGT-M and PGT-A from a single biopsy for a couple with *de novo* mutation in the CACNA1S gene without a reference. The combined analysis from a single embryo biopsy reduces the risk to the embryo, as well as optimizes the workflow of PGT in ART.