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The use of whole exome sequencing to identify carriers of multiple variegated aneuploidy (MVA): A case study

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Introduction: Whole-exome sequencing (WES) is a clinical diagnostic and research tool designed to identify and classify the arrangement of all protein-coding nuclear genes in the genome. Cases of infertility and recurrent miscarriages complicated by multiple-loop consanguinity may benefit from the implementation of whole-exome sequencing.

Case Presentation: We report a case of a Kuwaiti consanguineous couple referred to the genetics department with a history of three live births followed by fourteen miscarriages, most of which occurred during the third and fourth months of gestation. The initial routine investigations including the blood tests, thyroid function, imaging, immunological and antibody profile, and chromosomal microarray of both parents were normal. The presence of multiple loops of consanguinity made making a correct diagnosis difficult. The option of whole-exome sequencing to examine the entire exome of both parents was proposed. After taking the patients' consent, the WES results of the couple revealed them both to be carriers of a previously unreported heterozygous variant of the CEP57 mutation (HET CEP57 c.56del (p.Ala19Valfs*rtfaQN 19) resulting in multiple pregnancies with type 2 multiple variegated aneuploidy.

Methods: Whole-exome sequencing was agreed by the treating clinical geneticists as a suitable option to identify the presence of a possible genetic mutation. The age, gender, ethnicity, medical history, family history, consanguinity, laboratory investigation results, clinical interpretation and implication of the results have been documented.

Conclusion: The reported case of infertility and recurrent miscarriages associated with multiple-loop consanguinity showed no abnormalities using conventional diagnostic methods. The application of whole-exome sequencing for the couple revealed them to be carriers for the type 2 multiple variegated aneuploidy syndrome gene CEP57. In cases of multiple-loop consanguinity, whole-exome sequencing should be considered to aid in the diagnosis of the genetic mutations.

Biography

Munirah T Aljaser is a medical graduate of Kuwait University. A self-motivated and responsible individual, currently working as a Resident, treating patients at Al-Farwaniya Hospital.

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