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Allele amplification failure in the *HBB* gene due to allelic dropout in a pre-preimplantation genetic testing case

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Beta-Thalassemia is one of the common autosomal recessive inherited genetic conditions in the GCC region due to increased rates of consanguinity. It is a condition caused by several different types of mutations in the *HBB* gene. The current case represents a 30-year old gentleman who was informed to be a heterozygous carrier of β-thalassemia. At the clinic, peripheral blood was drawn for DNA sequencing. Although the patient indicated that he was a carrier, sequencing results surprisingly showed him to be affected with the codon 39 mutation which does not correspond at the genotype-phenotype level indicating the possibility of an allele dropout. Further analysis was carried out using two different sets of designed primers to confirm the carrier status. This showed that the gentleman is indeed a carrier of β-thalassemia at codon 39 of the *HBB* gene. In conclusion, to overcome and minimize the incidence of an allele dropout event in the pre-PGT setting and during PGT-M, it is advisable to detect every mutation using at least two different sets of primers to avoid unknown SNPs around the mutation region.

Biography

Mariam Fida has recieved her PhD in Medical Genetics from the Univesrity of Edinburgh, Scotland. She is also specialized in Preimplantation Genetics Diagnosis at Reproductive Genetic Innovations in Chicago. She established the first PGD center in Bahrain and is currently the Director of the Center and is a Consultant in Medical Genetics.

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