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Case study on identification of compound heterozygous β+ β0 mutation of *HBB* gene leading to β-thalassemia major in India

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β-thalassemia is a genetic disease characterized by reduced or non-functionality of *β-globin* gene expression, which is caused due to number of genetic variations and indels (insertions and deletions). In the present case study, we have reported a rare occurrence of compound heterozygosity of two different variants, namely, *HBB*c.92G>C and *HBB*c.92+5G>C in maternal amniotic fluid sample. Prenatal β-thalassemia mutation was detected using nucleotide sequencing method. After analysis, the father was found to be heterozygous for *HBB*c.92G>C (Codon 30 (G>C)) mutation (β0 type) and the mother was heterozygous for *HBB*c.92+5GNC (IVS I-5 (G>C)) mutation (β+ type). When amniotic fluid sample was analyzed for β- globin gene (*HBB*), we found the occurrence of heterozygous allelic pattern for aforesaid mutations. This compound heterozygous state of fetus sample was considered as β+/β0 category of β thalassemia which was clinically and genotypically interpreted as β-thalassemia major. The probability of occurrence of both mutations is very low, because mutations are only 5 base pairs apart on *HBB* gene. Segregation of compound heterozygosity has occurred twice in this family. Along with the present case, we will share our experience of analyzing 21 unrelated families (trios samples) for detection of β-thalassemia using whole gene sequencing and RT-PCR assays. We will share few interesting case studies like co-inheritance of sickle cell anemia and β-thalassemia traits, compound heterozygosity of beta thalassemia major and normal in the case of twin pregnancy. Prenatal diagnosis helps the parents to know the thalassemic status of the fetus in the first trimester screening.

Biography

Spandan Chaudhary is the Team Leader of Medical Genetics, Diagnostics and Next Generation research divisions of Xcelris Labs, India. He has six years of professional experience in genomics industry specifically in the Medical Genetics segment. He has developed more than 30 very important diagnostic, sports and nutrition health related assays. He has designed the beta thalassemia mutation screening assay based on whole gene sequencing of *HBB* gene including all the mutations and indels. He has screened more than 200 individual samples and 25 trio samples as prenatal screening for beta thalassemia. This approach is very useful in diagnosing prenatal thalassemia in combination with regular screening methods.

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