

Novel genomic variants associated with polysplenia, situs inversus totalis, atrial septal defect and double outlet right ventricle in Saudi patient

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Abstract

In this case study, we reported a case of 8-year-old Saudi patient diagnosed with polysplenia, situs inversus totalis and double outlet right ventricle. We identified five novel missense mutation in three genes GATA4, NIPBL and APC as causative mutations and could be used for early detecting of polysplenia syndrome.

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