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

Yousef Mohammedrabaa Hawsawi[1], Turki Mohammed Sobahy Sobahy[1], Samar Zailaie[1], Mohammed Baghdadi [1], Donya Bahussain[1], Othman Al-Zahrani[2], Chellasamy Panneerselvam[2], Edward Cupler[1], and Amjad Al-Kouatli[1]

[1]King Faisal Specialist Hospital and Research Centre - Jeddah
[2]University of Tabouk Faculty of Science

April 27, 2020

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.