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A Rare Case of Ostium Secundum ASD with AV Nodal Block in a Young Patient

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Abstract: ***Ostium primum ASD may be associated with high degree AV block largely due to the location of the defect and proximity of the AV node and His bundle to the defect. Ostium secundum ASD is very rarely associated with AV nodal block. Our patient presented with such a rare combination at a younger age, which prompted us to evaluate further towards association of any genetic syndromes. We are presenting this case for the rarity of the presentation.***

Keywords: Ostium primum ASD, AV block, KAT 6B, GENITO - PATELLAR SYNDROME, Hearing Loss

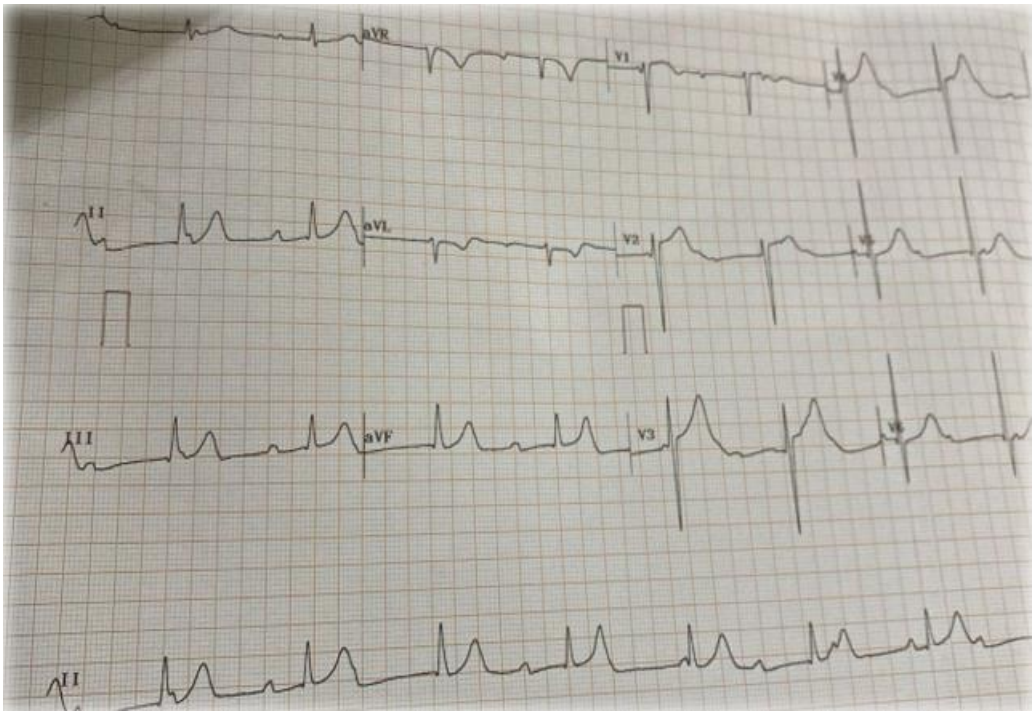
1. Case Report

Mrs. B, 30 - year - old female presented with chest pain, shortness of breath & giddiness for 2 days. O/E, patient was short, had pes planus and had low IQ. She also had hearing loss and irregular menstrual cycles. On CVS examination, she had an ejection systolic murmur with a loud P2.

Echocardiography showed ostium secundum ASD measuring 17mm with left to right shunt and a mild pulmonary hypertension. Postero - inferior rim was 1.6cm, antero - superior rim was 0.5cm and postero - inferior rim was insufficient. Basic investigations were normal. Interestingly, ECG showed complete heart block with supra - hisian escape with a rate of 50/min.

Literature review indicated that certain genetic syndromes were associated with a combination of ostium secundum ASD and complete heart block. Our patient also had certain general examination features which pointed towards a genetic predisposition.

We did a targeted genetic sequencing for the patient. It showed positive for KAT6B which is indicative for genitopatellar syndrome (OMIM#606170).



S. No	Gene, Variant details	Zygosity Depth (Alt allele %)	In silico tools	MAF	Literature	OMIM Disease	Inheritance	Classification
1	*KAT6B* (+) c.1489C>G p.Pro497Ala ENST00000287239.10	Heterozygous 112X(53.6%)	PolyPhen - BN MT2 - BN	gnomAD (V2.1) - 0.0003% gnomAD (V3.1) - 0.0006%	-	Genitopatellar syndrome (OMIM#606170)	Autosomal dominant	Uncertain Significance

2. Discussion

Genito patellar syndrome is an Autosomal dominant disorder characterized by short statue, small or absent patella, intellectual disability, cryptorchidism, club foot, microcephaly, agenesis of corpus callosum, micrognathia and hydroureteronephrosis, delay in eruption of teeth, hearing loss and thyroid issues. Cardiac manifestations include ostium secundum ASD and varying degrees of AV blockage including complete heart block. Our patient was short (140cm), had microcephaly, micrognathia and delayed eruption of teeth. She had menstrual irregularities (on treatment with a gynaecologist) a low IQ (confirmed by Psychiatry evaluation) & hearing loss authenticated by ENT examination. She presented with ostium secundum ASD and complete heart block and diagnosis was confirmed by a targeted genetic sequencing. We are presenting this case for the rarity of the presentation. We are planning ASD closure for the patient with further follow - up.

References

[1] Gessner IH. Atrial septal defect. In: Moller JH, ed. *Surgery of Congenital Heart Disease: Pediatric Care Consortium 1984 - 1995*. Armonk, NY: Futura Publishing Co.; 1998.31 - 44.

[2] Vick GW, Titus JL. Defects of the atrial septum, including the atrioventricular canal. IN: Garson A, Bricker JT, Fisher, DJ, Neish SR, eds. *The Science and Practice of Pediatric Cardiology*. Baltimore, MD: Lippincott Williams & Wilkins; 1998. Vol 2:

[3] Azhari N, Shihata MS, Al - Fatani A. Spontaneous closure of atrial septal defects within the oval fossa. *Cardiol Young*.2004 Apr.14 (2): 148 - 55. [QxMD MEDLINE Link].

[4] Saxena A, Divekar A, Soni NR. Natural history of secundum atrial septal defect revisited in the era of transcatheter closure. *Indian Heart J*.2005 Jan - Feb.57 (1): 35 - 8. [QxMD MEDLINE Link].

[5] Burn J, Brennan P, Little J, Holloway S, Coffey R, Somerville J, Dennis NR, Allan L, Arnold R, Deanfield JE, Godman M, Houston A, Keeton B, Oakley C, Scott O, Silove E, Wilkinson J, Pembrey M, Hunter AS. Recurrence risks in offspring of adults with major heart defects: results from first cohort of British collaborative study. *Lancet.1*998; *351*: 311–316.