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Nonsurgical device closure of isolated ostium secundum atrial septal defect in a case of genetically proven tuberous sclerosis (TSC2)

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Tuberous sclerosis (TS) is an autosomal dominant multisystem disease associated with multiple cardiac rhabdomyomas; but congenital cardiac malformations are very rare in TS. Two genes implicated in tuberous sclerosis have been identified which are the tuberous sclerosis 1 (TSC1) hamartin gene on chromosome 9 (9q34) and the tuberous sclerosis 2 (TSC2) tuberin gene on chromosome 16 (16p13.3).

A five-year-old girl with classical features of TS had an associated large secundum atrial septal defect (ASD). The left to right shunt through the ASD was augmented by the multiple left ventricular masses. Genetic analysis showed heterozygous 3" splice site variation in intron 3 of TSC2 gene (chr16:2103342; G>G/A; Depth: 71x) that affects the invariant GA acceptor slice site of exon 4 (c.226-1G>G/A; ENST00000219476). Non-surgical closure of the ASD avoided the neurological and cardiac complications that may occur due to cardiopulmonary bypass during open-heart surgery.

This first novel interventional report on TS stresses on the value of such catheter interventions to mitigate risks in complex cardiac associations.