Persistent Central Carpal Row Ossicle in a Sporadic Case of Holt-Oram Syndrome

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Background / Holt – Oram syndrome (HOS)

-incidence 0.7-1 / 100,000 births
-autosomal dominant inheritance, caused by mutations in TBX5 gene *(12p24.1)*
->70% of patients have identifiable mutations in the TBX5 gene
- 100% penetrance but variable expression, especially for the upper extremity
-skeletal upper extremity malformations severity F>M, L>R

Case presentation

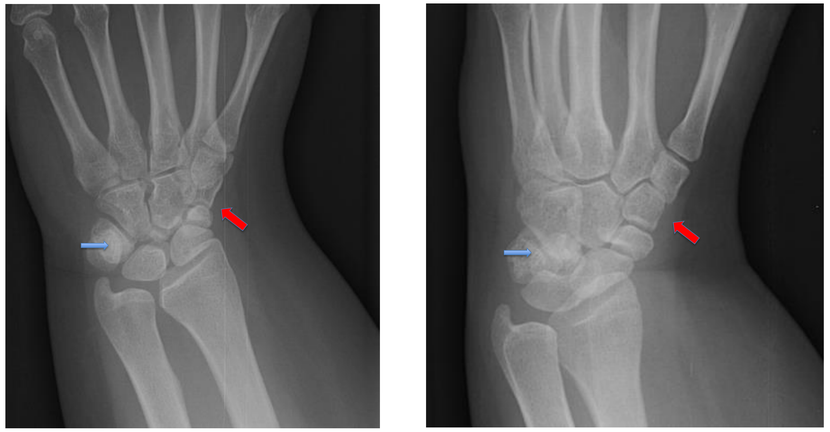
20 yo F, presents complaining of a 2 month history of worsening left wrist pain.

PMH: complete heart block, dual chamber pacer, small asymptomatic ventral septal defect
PSH: none
FH: Negative for cardiac abnormalities, upper extremity deformities, no diagnosis of HOS

Physical Exam findings

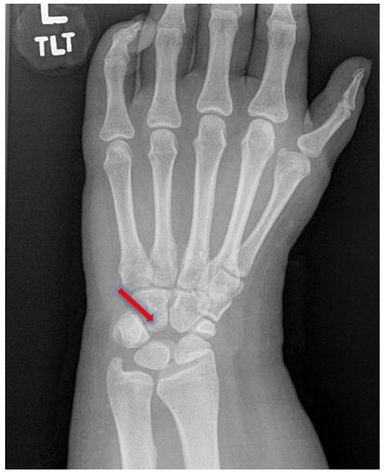
-hypoplastic, mildly shortened L thumb with increased MP laxity (*Blauth type 2*)
-pain on supination, radial deviation, axial wrist loading
-R hand normal on exam

Pre-op imaging findings



-**Supernumerary radial carpal bones between scaphoid and trapezium**
-**Persistent central row ossicle (*os centrale*) articulating with capitate / hamate / scaphoid / lunate**

Management-outcome



-surgical exploration
-articular surfaces of scaphoid, lunate, capitate and hammate intact
-*os centrale* completely excised
-complete symptom resolution at 3 month follow up

Cardiac manifestations

-75% of patients have structural and conduction anomalies
-*ostium secundum* ASD is the most defect followed by VSD
-1st degree block is the most common conduction anomaly

Upper extremity manifestations

-significant phenotypic variability
-phenotypes consistent with radial longitudinally deficiency
-absent or hypoplastic thumb is common

Carpal manifestations

-very specific, found in *ALL* affected individuals
-length deficient scaphoid with interposing bones between scaphoid and trapezium
-the *os centrale,* a remnant of the central carpal row can be rarely preserved, usually partially fused to the scaphoid

Conclusions

-although under-reported, carpal bone abnormalities are the most specific phenotype for the syndrome

-carpal bone abnormalities should raise suspicion for undiagnosed, sporadic Holt-Oram syndrome

-patients should be offered genetic testing /counseling upon Holt-Oram syndrome suspicion

-formal cardiac evaluation is necessary before planning any surgical intervention

References: McDermott DA et al. Holt-Oram syndrome. GeneReviews®, University of Washington, Seattle; 1993-2015 / Wall LB, et al. Defining features of the upper extremity in Holt-Oram syndrome. J Hand Surg Am. 2015 Sep;40(9):1764-8 / Poznanski AK et al. Skeletal Manifestations in the Holt-Oram syndrome. Radiology. 1970 Jan;94(1):45-53