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 
P>F>M, L>R

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

References:

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 hamamate intact
- os centrale completely excised
- complete symptom resolution at 3 month follow up

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