

# C-42

## Currarino Syndrome: A Rare Condition with Potential Connection to Neuroendocrine Tumors



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**BACKGROUND:** Currarino syndrome (CS) is a congenital disorder that consists of a triad of anomalies: presacral mass, sacral dysgenesis, and anorectal malformations. It is an autosomal dominant condition that has been associated with mutations in the MNX1 gene. There have been few reports in the literature regarding the association of CS with neuroendocrine tumors (NETs). In this study, we sought to determine the incidence and clinical characteristics of NET in patients with CS.

**METHODS:** Mayo Clinic electronic medical records were searched for patients with CS. Data on demographics, CS diagnosis (made clinically or genetically), NET diagnosis, family history of NET, were extracted.

**RESULTS:** A total of 26 patients were identified. Genetic testing was confirmed in 11/26 patients. Only 3/26 (11.5%) of patients were found to have a concomitant diagnosis of NET with ages ranging from 22 years to 75 years at time of NET diagnosis. 3/26 patients had family history of NET. Out of the three patients with CS and NET, two had confirmed primary NET from the pre-sacral mass. The remaining patient demonstrated focal uptake within a pre-sacral mass with metastatic liver lesions, currently pending surgical evaluation for pre-sacral mass resection. Two patients received octreotide and then subsequent peptide receptor radionuclide therapy due to progression of the metastatic disease. The third patient was not treated due to complete resection of pre-sacral mass with negative margins, and currently undergoing surveillance scans.

**CONCLUSION:** In our cohort of patients with Currarino syndrome, the incidence of neuroendocrine tumors is estimated at 11.53%. The coexistence of two very rare conditions, CS and presacral primary NETs and the absence of association with NETs arising in other locations, suggests there may be an etiological connection between CS and presacral primary NETs.