

Teaching NeuroImages: Short stature, imperforate anus, and polydactyly

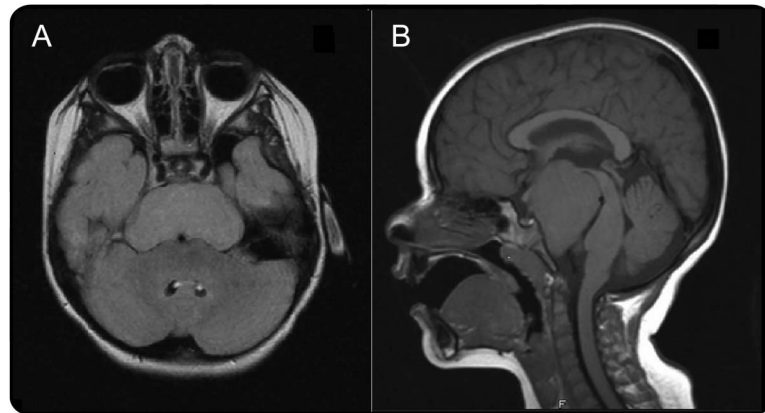
When is a hypothalamic mass an incidentaloma?

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Figure Imaging features of a large hypothalamic hamartoma



Brain MRI in axial (A) and sagittal (B) planes shows a large homogeneous mass (approximately $3.5 \times 5 \times 6$ cm) in the preoptic cistern, deriving from hypothalamus. There is stretching anteriorly of the optic chiasm and posteriorly there is mass effect on the midbrain and pons, with no parenchymal edema.

A 4-year-old girl with history of polydactyly and imperforate anus surgeries presented with short stature. Brain MRI revealed a large hypothalamic hamartoma (figure). Pallister-Hall syndrome (PHS) was diagnosed. Biopsy was not indicated given stable and typical tumoral appearance. Annual evaluations, hormonal replacement, and medical genetics consult were recommended. Compared to sporadic hypothalamic hamartoma, patients with PHS tend to be less symptomatic and have minimal tumor growth.¹ The etiology is an autosomal dominant or de novo *GLI3* gene mutation.² Neurologists and neuroradiologists should be familiar with PHS characteristics, surveillance, and symptomatic therapy as neurosurgery is not usually recommended.

AUTHOR CONTRIBUTIONS

Oana M. Dumitrascu: drafting/ revising the manuscript, study concept or design, analysis or interpretation of data, accepts responsibility for conduct of research and final approval. Patrick Lyden: drafting/ revising the

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DISCLOSURE

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