Short stature, imperforate anus, and polydactyly
When is a hypothalamic mass an incidentaloma?

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. The etiology is an autosomal dominant or de novo GLI3 gene mutation. 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 manuscript, study concept or design, analysis or interpretation of data, accepts responsibility for conduct of research and final approval, study supervision. Moise Danielpour: drafting/revising the manuscript, study concept or design, analysis or interpretation of data, accepts responsibility for conduct of research and final approval, study supervision. Franklin Moser: drafting/revising the manuscript, study concept or design, analysis or interpretation of data, accepts responsibility for conduct of research and final approval, study supervision.

STUDY FUNDING
No targeted funding reported.

DISCLOSURE
The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

REFERENCES
Teaching NeuroImages: Short stature, imperforate anus, and polydactyly: When is a hypothalamic mass an incidentaloma?
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Neurology 2015;84:e117
DOI 10.1212/WNL.0000000000001469

This information is current as of April 13, 2015

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