RESIDENT & FELLOW SECTION

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Teaching NeuroImages: Isolated hypothalamic hamartoma vs Pallister-Hall syndrome

Imaging and clinical correlation

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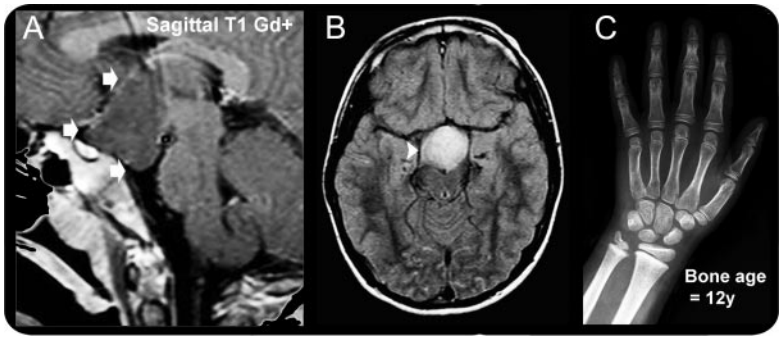
Isolated hypothalamic hamartomas (HH) have a distinct clinical phenotype from that of Pallister-Hall syndrome (PHS), as HH consist of more severe seizures and cognitive, behavioral, and endocrine disorders.[1] The imaging features help one to distinguish these conditions (figures 1 and 2).

Isolated HH are hyperintense on fluid-attenuated inversion recovery and cause precocious puberty (PP) when they are oriented downward (parahypothalamic lesion), while seizures predominate in sessile intrahypothalamic HH.[2] PHS is associated with isointense to gray matter HH, polydactyly, cutaneous syndactyly, bifid epiglottis, imperforate anus, and panhypopituitarism, resulting from GLI3 frameshift mutations (which map to chromosome 7p13) as an autosomal dominant trait.[1]

AUTHOR CONTRIBUTIONS

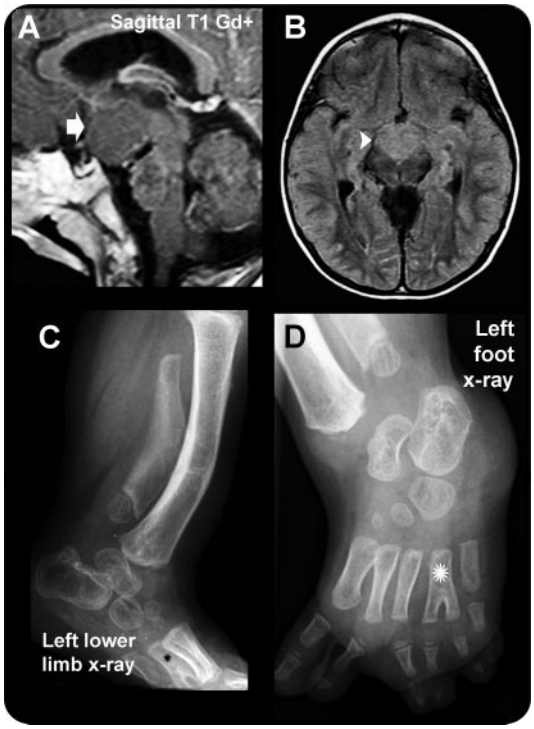
Dr. Rocha was responsible for the study concept, writing the manuscript, and the interpretation of data. Dr. Rosa Jr. was responsible for the interpretation of data and image selection. Dr. Arita was responsible for the clinical study concept and critical revision of the manuscript.

Figure 1 Isolated hypothalamic hamartoma (HH) (3-year-old boy) with gelastic epilepsy and precocious puberty



(A, B) MRI confirmed nonenhancing HH (arrows). Typical hyperintensity on fluid-attenuated inversion recovery (arrowhead). (C) Advanced bone age.

Figure 2 Pallister-Hall syndrome (2-year-old boy)



(A, B) MRI confirmed nonenhancing hypothalamic hamartoma (arrow) that was isointense to gray matter on fluid-attenuated inversion recovery (arrowhead). (C, D) Note the curved long bones, polydactyly, and dysplastic metatarsal (asterisks).

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MYSTERY CASE RESPONSES

The Mystery Case series was initiated by the *Neurology*® Resident & Fellow Section to develop the clinical reasoning skills of trainees. Residency programs, medical student preceptors, and individuals were in-

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vited to use this Mystery Case as an education tool. Responses were solicited through a group e-mail sent to the AAN Consortium of Neurology Residents and Fellows and through social media.

All the answers that we received came from individual residents rather than groups and they were all well-reasoned and thoughtful. Three respondents correctly identified the classic appearance of isolated hypothalamic hamartomas (figure 1) and Pallister-Hall syndrome (figure 2). When isolated, hypothalamic hamartomas present with severe epilepsy, more significant neurologic dysfunction, and are more likely to be associated with precocious puberty. Patients with Pallister-Hall syndrome usually have well-controlled seizures and endocrine disturbances other than precocious puberty.

The teaching point of this Mystery Case is that despite similar appearance the exact MRI localization and the T2/fluid-attenuated inversion recovery characteristics may help in differentiating between these 2 entities, and thus the imaging could be used as a prognostic factor for the clinical evolution.

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Neurology 2012;79;950-951
DOI 10.1212/WNL.0b013e3182676796