Medulloblastoma associated with novel \textit{PTCH} mutation as primary manifestation of Gorlin syndrome

A 3-year-old boy with a history of developmental delay presented with a 2-week history of vomiting and irritability. Neurologic examination revealed an encephalopathic child without focal deficits. Parafalcine nodular calcifications on head CT (figure, B) in association with hypogenetic corpus callosum on MRI (figure, C) and desmoplastic medulloblastoma pathology (figure, D) raised suspicion for basal cell carcinoma nevus syndrome (Gorlin syndrome).\textsuperscript{1} Falcine calcifications are rare in children and can be associated with trauma, infection, meningiomas, and pseudohypoparathyroidism. \textit{PTCH} tumor suppressor gene mutation analysis demonstrated a novel Y601X nonsense mutation in exon 13. Based on the genetic findings, radiation therapy was withheld in our patient due to the risks of secondary malignancies in patients with Gorlin syndrome.\textsuperscript{2}

John R. Crawford, MD, MS, Brian R. Rood, MD, Christopher T. Rossi, MD, Gilbert Vezina, MD, Washington, DC

Disclosure: The authors report no disclosures.

Address correspondence and reprint requests to Dr. John R. Crawford, Department of Neurology, Children’s National Medical Center, The George Washington University, Washington, DC 20010; jcrawfo@cnmc.org


Medulloblastoma associated with novel PTCH mutation as primary manifestation of Gorlin syndrome
Neurology 2009;72;1618
DOI 10.1212/WNL.0b013e3181a413d6

This information is current as of May 4, 2009

Updated Information & Services
including high resolution figures, can be found at:
http://n.neurology.org/content/72/18/1618.full

References
This article cites 2 articles, 0 of which you can access for free at:
http://n.neurology.org/content/72/18/1618.full#ref-list-1

Subspecialty Collections
This article, along with others on similar topics, appears in the following collection(s):
Primary brain tumor
http://n.neurology.org/cgi/collection/primary_brain_tumor

Permissions & Licensing
Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at:
http://www.neurology.org/about/about_the_journal#permissions

Reprints
Information about ordering reprints can be found online:
http://n.neurology.org/subscribers/advertise