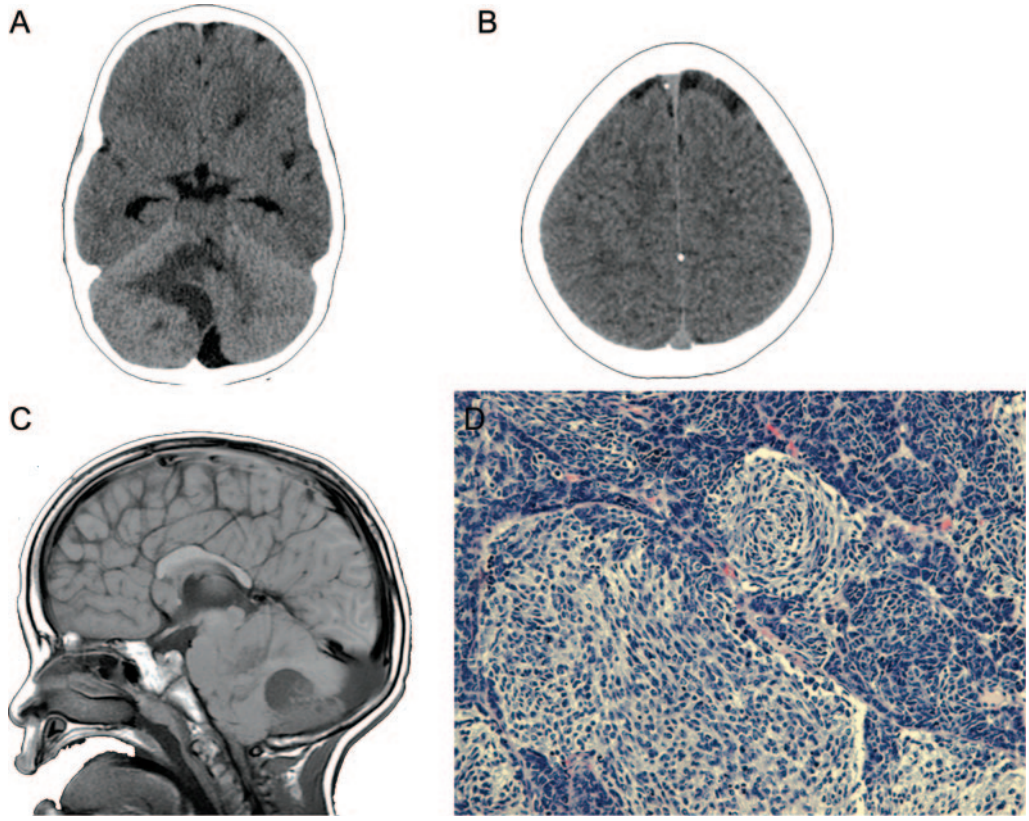
Medulloblastoma associated with novel *PTCH* mutation as primary manifestation of Gorlin syndrome

Figure Head CT, MRI, and tumor pathology



(A) Noncontrast head CT shows a posterior fossa mass with associated hydrocephalus and parafalcine nodular calcifications (B). (C) Sagittal T1 noncontrast MRI further reveals macrocephaly and hypogenetic corpus callosum. (D) Hematoxylin & eosin–stained tumor specimen demonstrates the reticular-nodular appearance of desmoplastic medulloblastoma.

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).[1] Falcine calcifications are rare in children and can be associated with trauma, infection, meningiomas, and pseudohypoparathyroidism. *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.[2]

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

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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; jcrawfor@cnmc.org

1. Kimonis VE, Mehta SG, Digiovanna JJ, Bale SJ, Pastakia B. Radiological features in 82 patients with nevoid basal cell carcinoma (NBCC or Gorlin) syndrome. Genet Med 2004;6:495–502.
2. Epstein EH. Basal cell carcinomas: attack of the hedgehog. Nat Rev Cancer 2008;10:743–754.

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