Medulloblastoma associated with novel *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). 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.

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Disclosure: The authors report no disclosures.

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Neurology 2009;72;1618
DOI 10.1212/WNL.0b013e3181a413d6

This information is current as of May 4, 2009