Multiple nevoid basal-cell carcinoma syndrome (Gorlin's syndrome): Possible confusion with metastatic medulloblastoma

Case report

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The authors report a diagnostic dilemma involving a child who, 8 years previously, had total excision of a medulloblastoma. On x-ray studies, lytic lesions of the skull were seen. The differential diagnosis and some of the clinical and pathological aspects of the nevoid basal-cell carcinoma syndrome versus metastases are discussed.

KEY WORDS: bone cyst, dural calcification, Gorlin's syndrome, medulloblastoma, nevoid basal-cell carcinoma

Gorlin's syndrome is primarily characterized by multiple nevoid basal-cell carcinomas (NBCC), jaw cysts, skeletal anomalies, and a positive family history. There are many associated lesions reported. Some prefer to classify these cases as the NBCC syndrome, which has the following cardinal features: "(1) Multiple nevoid cutaneous basal cell carcinoma, (2) cysts of the jaw, (3) rib anomalies, (4) pits on the hands and soles and (5) intracranial calcification." Others use the term NBCC syndrome to denote basal-cell nevi, broad faces, and rib anomalies. Probably these are variable expressions of the same disorder. What they have in common is important to the neurosurgeon, especially the pediatric neurosurgeon, since medulloblastoma is known to occur with both.

Case Report

In September, 1968, this baby boy, aged 2½ years, was admitted to The Hospital for Sick Children, Toronto, and underwent excision of a left cerebellar medulloblastoma. One week before the removal, he had a right ventriculoperitoneal shunt inserted. Two weeks postoperatively he began a 6-week course of radiotherapy. During this time he received 5263 rads to the skull and 4008 rads to the spine. Postoperatively, no neurological deficit could be demonstrated. Five years later, in November, 1973, a non-pigmented lesion of his forehead was biopsied. Microscopic examination was consistent with basal-cell carcinoma showing some adenocystic changes.

In March, 1977, when the boy was aged 11 years, a dental x-ray examination revealed cystic lesions of the mandible and lytic lesions of the skull. The lesions of the mandible were resected and the microscopic examination showed an epithelial-lined cystic lesion compatible with those seen in Gorlin's syndrome. At this time, the patient was referred back to The Hospital for Sick Children. There was no positive family history of the NBCC syndrome.

Examination. The patient was markedly obese with a round, jowled face. Several nevi were distributed over the upper trunk and neck. There were pinpoint dyskeratotic lesions of both palms. His testes were soft and atrophic, with a small penis buried in suprapubic fat. His skull was characterized by frontal and parietal bossing. Hypertelorism was borderline. He had no neurological deficit.

On skull films (Fig. 1), multiple lytic lesions, 3 to 4 mm in size, were noted, as well as calcification of the falx, neither of which had been seen on prior films. The lytic lesions were consistent with metastatic disease. Cysts of the mandible and multiple vertebral anomalies in the thoracic area were also seen.

Radionuclide bone scan of the skull (Fig. 2)
Nevoid basal-cell carcinoma of the skull

Operation. Since no evidence had been obtained to confirm that this lad had metastatic disease, a biopsy of the left parietal region was carried out on March 17, 1977. At operation, the skin and periosteum were

![Image 1](image1.png)

**Fig. 1.** Lateral and anteroposterior skull films taken in March, 1977, with multiple lytic areas. The burr hole for the shunting system is seen in the right parieto-occipital region. Note the calcification of the falx and the tentorium.

![Image 2](image2.png)

**Fig. 2.** Radionuclide bone scan of the skull in March, 1977, lateral and anteroposterior views, showing the multiple areas interpreted as metastatic lesions.
believed to be normal. The skull was firm but had a blotchy appearance with flat purplish lesions that extended from the outer to the inner table. The areas were not cystic on gross examination. No evidence of metastatic disease was noted but several focal areas of pseudocysts were found on microscopic examination.  

Postoperative Course. At his 8-month follow-up examination, the patient continued to do well. He had no neurological deficit, and no new bone lesions.

Discussion

The patient with a previous medulloblastoma and lytic lesions in the skeleton or skull should be suspected of having metastatic lesions.\textsuperscript{1,6,7} The authors believe that the likelihood of metastasis is increased when a shunting system is used before and during removal of the tumor, unless a millipore filter is incorporated into the cerebrospinal fluid bypass system. The system placed in this patient did not have a filter.\textsuperscript{8,7} The lesions of the skull were not unlike those previously reported with medulloblastoma. The radionuclide bone scan of the skull also suggested metastatic lesions.

Caldwell reported a patient with follicular cysts of the maxilla and mandible,\textsuperscript{2} which are recognized as an integral part of the multiple NBCC syndrome. The skull film revealed a “woolly hair” appearance, and x-ray films of the femurs revealed punched-out lesions.\textsuperscript{2} In our patient, the “woolly hair” appearance was not present, but lesions not dissimilar to the reported femoral lesions\textsuperscript{2} were found in the skull. The decision to biopsy despite firm assurance from our radiographers that these were metastatic lesions was based on our dictum that tissue must be obtained in all lesions before radiation therapy.

In 1971, Gorlin\textsuperscript{3} reviewed more than 100 reports of this syndrome and found that the chief components were multiple NBCC, odontogenic keratocysts of the jaws, various skeletal anomalies, a characteristic facies with frontal and temporoparietal bossing and well developed supraorbital ridges, intracranial calcification, primarily of the dura, medulloblastoma, ovarian fibromas, and lymphomesenteric cysts. He noted that the syndrome is likely “autosomal dominant with high penetrance and variable expressivity.”\textsuperscript{4} It was suggested that the association with medulloblastoma is higher, but before modern surgical techniques, radiation, and chemotherapy, these patients succumbed before the other stigmata of the syndrome developed.

The relative frequency of the central nervous system (CNS) involvement, in particular, calcification of the falx, equals that of the multiple jaw cysts, mild mandibular prognathism, dystopia canthorum, hypertelorism, ovarian fibromas, or cysts, and the multiple NBCC.\textsuperscript{5}

The aspects of the CNS that may be involved are, in order of decreasing frequency: calcification of the dura, falx, or tentorium; choroid; petroclinoid liga-

ment; mental retardation of varying degrees, schizophrenia; congenital hydrocephalus; agenesis of the corpus callosum; medulloblastoma; and nerve deafness.\textsuperscript{6}

Neblett, et al.,\textsuperscript{8} reported four cases of medulloblastoma with the multiple NBCC syndrome, and reviewed the literature. They stated that medulloblastomas seemed to appear earlier, noting that 50% of the cases were under 2 years of age when the diagnosis of medulloblastoma was made. Their article also suggests a longer survival time if the NBCC syndrome is established.

In summary, lytic skull lesions in the patient with medulloblastoma may be part of the Gorlin syndrome or a larger multiple NBCC syndrome and not metastatic lesions. This finding has apparent implication for the management of these patients.

References