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Gorlin Syndrome: Unusual Manifestations in the Sella Turcica and the Sphenoidal Sinus

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Summary: We report a case of Gorlin syndrome in a 20-year-old woman in whom CT and MR images showed unusual findings of the pituitary gland and the sphenoidal sinus, suggesting previously unrecognized lesions associated with this disease.

Nevoid basal cell carcinoma (Gorlin syndrome) classically consists of multiple basal cell carcinomas of the skin, odontogenic keratocysts of the jaw, various skeletal abnormalities, and lamellar falx calcifications (1). Many associated lesions have been reported (2–5). We present a case of this syndrome, with special emphasis on its unusual neuroradiologic findings.

Case Report

A 20-year-old woman presented with polymenorrhea occurring every 20 days. Physical examination revealed a prominent forehead, hypertelorism, mild exophthalmos, and several scattered pigmented lesions of the skin. Histologically, there was no evidence of malignant lesions of the skin. Laboratory examination of the pituitary function revealed normal levels of luteinizing hormone, follicle-stimulating hormone, growth hormone, and prolactin. There was no evidence of diabetes insipidus, and family history was unremarkable.

Skull radiographs and head CT scans showed extensive calcifications in the falx, in the tentorium, and in the diaphragma sellae (Fig 1A–C). Coronal CT scans showed the pneumatized lateral recess of the sphenoidal sinus interposing between the pituitary gland and the cavernous sinus (Fig 1C). A panoramic radiograph showed a cystic lesion in the body of the right mandible. An MR imaging examination, obtained to investigate the patient's mental retardation, revealed extensive pneumatization of the sphenoidal sinus and downward convexity of the sellar floor, best depicted on coronal images (Fig 1D). The pituitary gland was of normal volume but had an unusual rounded configuration. The pituitary gland was bounded laterally by the upper lateral recess of the sphenoidal sinus, not bounded by the cavernous sinus as seen on coronal CT scans. The rest of the sinuses were normally pneumatized. On sagittal MR images, a focal cystic structure was seen in the posterior aspect of the pituitary fossa, and there was loss of the usual posterior pituitary lobe bright spot (Fig 1E and F). Mild cerebral atrophy was visible, but there was no evidence of intracranial neoplasms. Other radiologic findings included a splayed appearance of the right fifth rib and a large calcified pelvic mass.

At abdominal laparotomy, a solid multinodular tumor was found, almost completely replacing the left ovary and measuring 8.5 × 8 × 7 cm; there were also two small solid tumors located on the surface of the right ovary. The tumors were excised and a left salpingo-oophrectomy was performed. The microscopic diagnosis was multiple fibromas of both ovaries. The patient's postoperative course was uneventful, but polymenorrhea was still present 6 months after laparotomy. The mandibular cyst was subsequently excised and diagnosed as a keratocyst.

Discussion

In 1960, Gorlin and Goltz described the association of basal cell carcinomas, odontogenic keratocysts, and bifid ribs (6). Since that time, a multitude of additional manifestations of Gorlin syndrome has been reported, including medulloblastoma or craniofacial dysmorphism (2–5). Gorlin syndrome shows an autosomal dominant pattern of inheritance with variable expressivity. Recent molecular genetic studies have suggested that a mutation of a tumor-suppressor gene may relate to the pathogenesis of Gorlin syndrome, including development of neoplasms or various anomalies (7, 8). In general, the diagnosis of Gorlin syndrome can be made when two or more of the following abnormalities are noted: onset of multiple nevoid basal cell carcinomas early in life, cysts of the jaw, palmar and plantar pits, ectopic lamellar calcification of the falx, and developmental skeletal anomalies (5, 9). Ovarian fibroma is also known to be frequently associated with Gorlin syndrome (9, 10). Although our patient did not have basal cell carcinomas of the skin, she did have a mandibular keratocyst, calcification of the falx, splayed rib, and ovarian fibromas, all of which suggested the diagnosis of Gorlin syndrome.

One of the most common intracranial abnormalities in Gorlin syndrome is lamellar calcification of the falx, which occurs in up to 67% of patients (5). Calcification has also been described in the tentorium, petroclinoid ligaments, dura, pia, choroid plexus, and basal ganglia.

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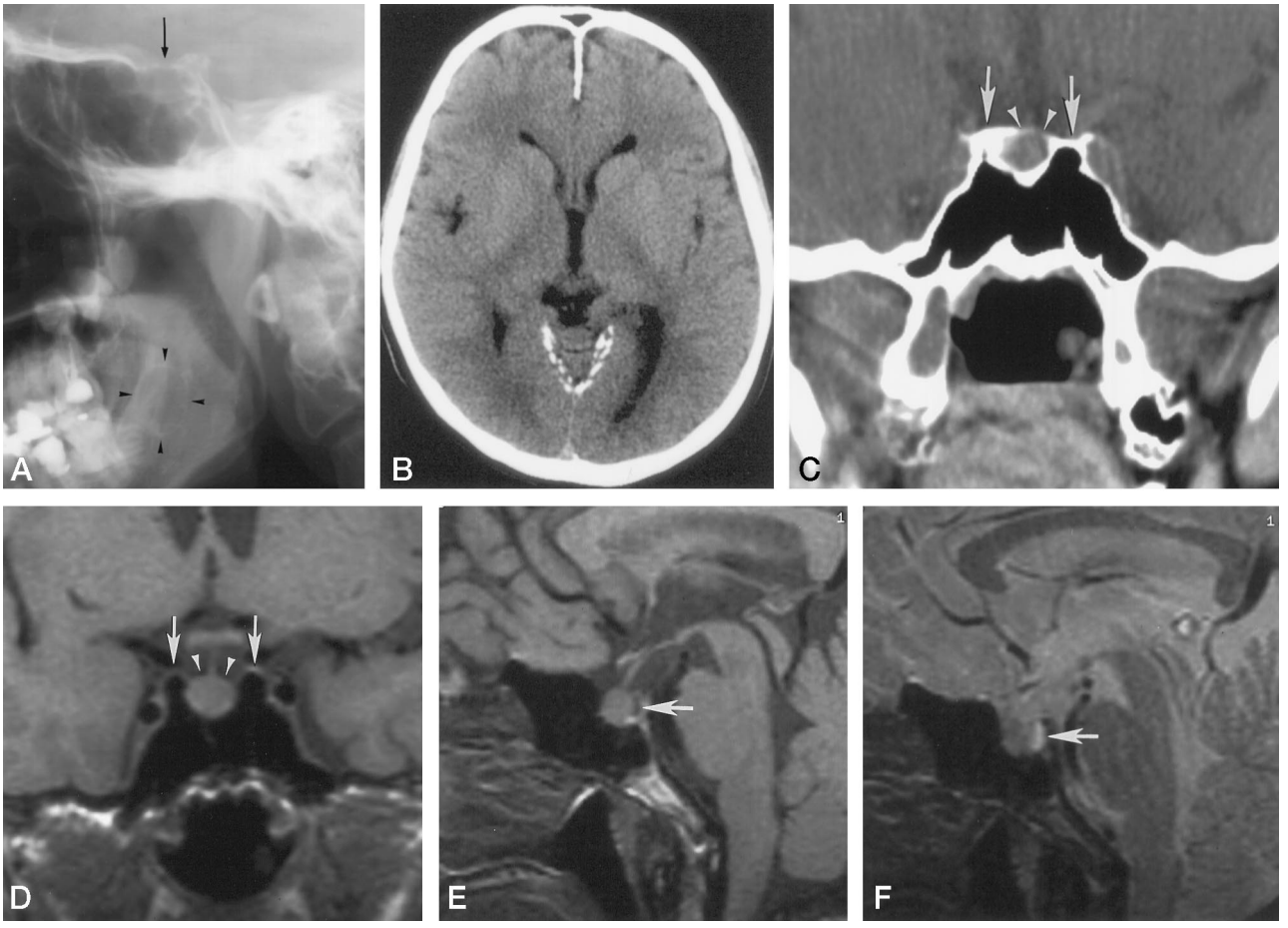

FIG 1. 20-year-old woman with nevoid basal cell carcinoma (Gorlin syndrome).

A, Lateral view of a skull radiograph shows calcification of the diaphragma sellae (*arrow*) and a mandibular cystic lesion (*arrowheads*).

B, Axial CT scan shows calcifications of the falx cerebri and the tentorium.

C, Coronal CT scan shows calcification of the diaphragma sellae (*arrowheads*). Note the lateral recess of the sphenoidal sinus extending superiorly and bounding the pituitary gland laterally (*arrow*).

D, Coronal T1-weighted (600/26/4) MR image shows downward convexity of the sellar floor. The pituitary gland reveals a rounded configuration and is bounded laterally by a markedly pneumatized sphenoidal sinus (*arrows*). There are low signal bands, representing calcifications of the diaphragma sellae, adjacent to the top of the pituitary gland (*arrowheads*).

E and *F*, Sagittal T1-weighted (600/26/4) and T2-weighted (2000/90/2) MR images, respectively, show the absence of the posterior pituitary bright spot and a focal cystic lesion of the posterior aspect of the pituitary fossa (*arrows*).

With regard to the abnormalities of the sella turcica associated with Gorlin syndrome, a shallow or bridging configuration has been reported (2, 4, 10). In healthy subjects, the sellar floor is usually flat or downwardly convex, and the maximum depth of the central depression measures up to 3.5 mm on a frontal tomogram (11). However, the sellar floor of our patient showed marked downward convexity (11 mm of central depression), with a spherically shaped pituitary gland. There was no evidence of pituitary dysfunction; however, the persistent polymenorrhea may indicate a potential anomaly of the pituitary gland. Additionally, a small cystic area was seen in the posterior aspect of the pituitary fossa, with absence of the normal posterior pituitary lobe bright spot. The exact cause of the absence of the bright spot in this case is unknown; however, an incidental small Rathke cleft cyst or other epithelial cystic lesion may have been responsible (12).

The markedly pneumatized lateral recess of the sphenoidal sinus interposing itself between the pituitary fossa and the cavernous sinus in this case is unique. Pneumatization of the sphenoidal sinus more commonly extends into anterior clinoid processes and medial pterygoid processes than into the dorsum sella and posterior clinoid processes (13, 14). Mosskin et al (8) described hyperpneumatization of the paranasal sinuses as a manifestation of Gorlin syndrome after reviewing the literature. However, to the best of our knowledge, there has been no detailed radiologic description of hyperpneumatization of the sphenoidal sinus associated with such unusual appearances of the sella turcica and pituitary gland. These findings may represent a new associated manifestation of Gorlin syndrome.

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