Images in Clinical Medicine: McCune-Albright Syndrome

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Twelve years ago, the patient on the left (Figure 1), one of the 29-year-old single-placenta monozygotic twin sisters, underwent computed tomography after a relatively mild head trauma. As an incidental finding, the computed tomography scan (Figure 2) revealed that the skull structure was consistent with fibrous dysplasia, a characteristic finding for McCune-Albright syndrome. The pituitary gland was slightly enlarged. The second twin had no features of McCune-Albright syndrome.

McCune-Albright syndrome is a sporadic disorder, caused by postzygotic activating mutations of the GNAS1 gene and characterized by polyostotic fibrous dysplasia, pigmented patches of skin, and endocrinological abnormalities (1). The extent and the nature of the abnormalities are highly variable, depending on the specific tissues involved in mosaicism.

The affected twin had regular menses beginning at age 13. Her zygoma was very prominent on the left side, a manifestation of fibrous dysplasia, and the facial structure was typical of acromegaly (broad nose, elongated jaw, large mandible, and sharp facial features). There was an oval-shaped hyperpigmented lesion on the lower chest. Laboratory tests revealed high levels of serum prolactin, IGF-1, and GH (without oral glucose challenge test stimulation), which led to a diagnosis of acromegaly. The levels of TSH and T4 were normal. Molecular analysis of the GNAS1 gene from both twins did not reveal the cause of the discordance. Treatment of acromegaly was initiated with a somatostatin analog and a dopamine agonist, and fibrous dysplasia was managed with bisphosphonates. Under this treatment, GH and IGF-1 stabilized at slightly increased levels. Over the years, the phenotypic features of acromegaly became more prominent.

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Reference

Figure 2. This multiplanar reformation cut was performed through superior parts of orbits and posterior part of foramen magnum. Either involvement of most frequent sphenoid and ethmoid (mostly left parts) or unusual involvement of temporal (left) and occipital bones is seen.