

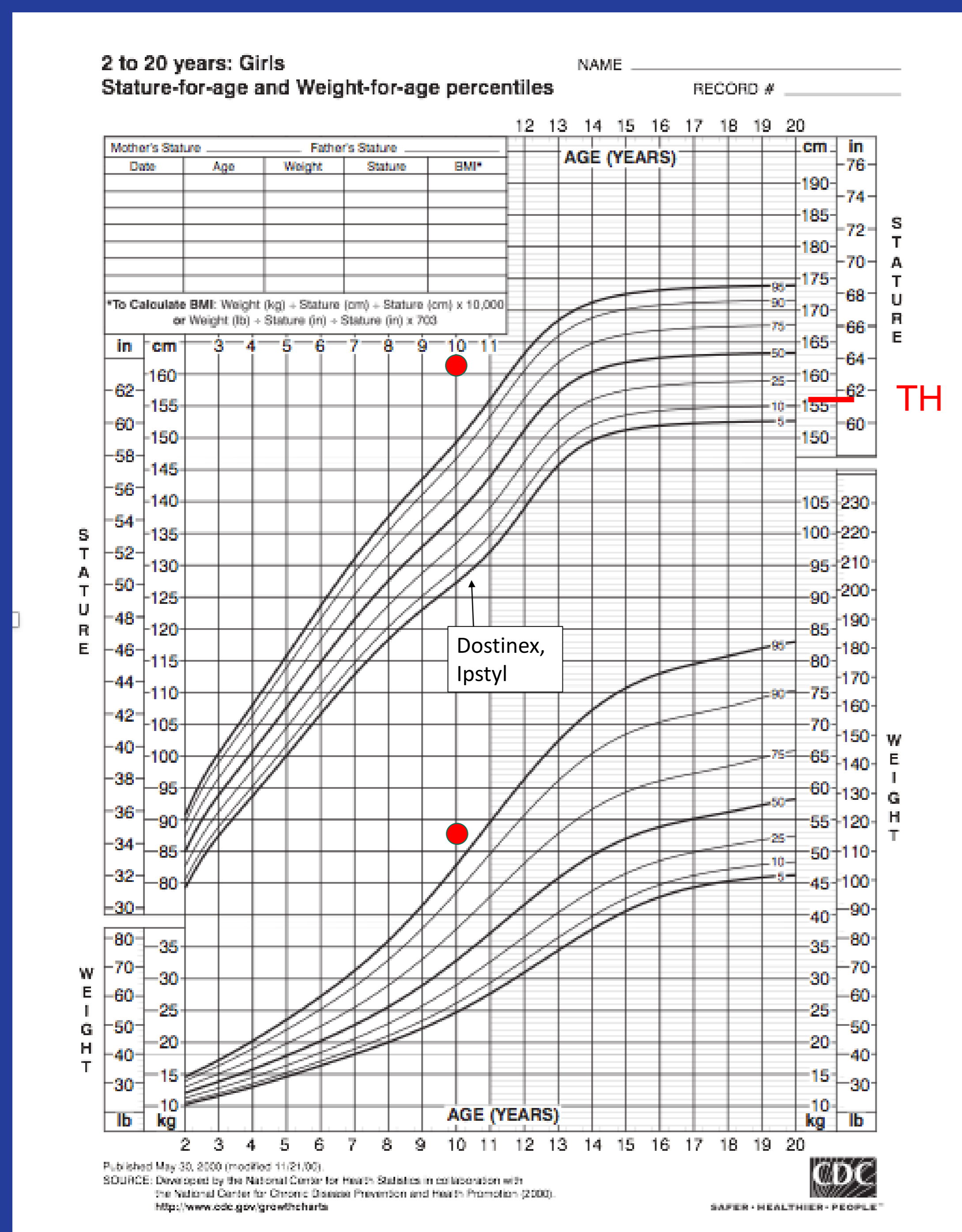
UNUSUAL PRESENTATION OF MCCUNE-ALBRIGHT SYNDROME IN A 10-YEAR-OLD GIRL

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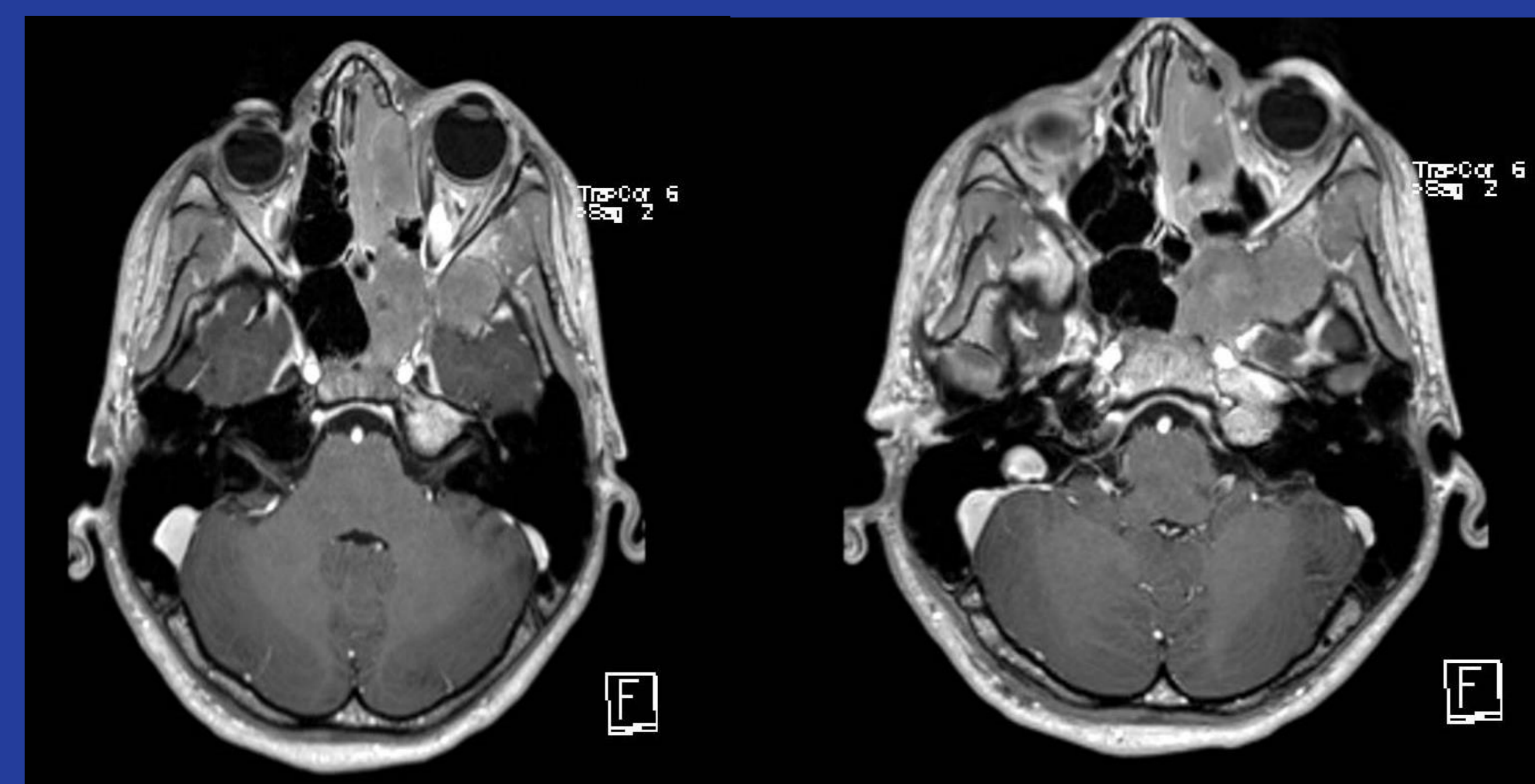


CASE REPORT



A 10-year-old Italian girl presented with a painless, hard swelling in left fronto-orbital region noted two years earlier. She had no symptoms; ophthalmologic evaluation revealed no signs of intracranial hypertension; the visual field showed a reduced level of sensitivity in the upper sector of the left eye. The girl was a second born after an uneventful pregnancy with normal delivery from healthy, unrelated parents. Birth weight was 3700 g, psychomotor development had been regular and no previous hospital admissions were reported.

At clinical examination, the child was in good general condition. Neurologic examination was unremarkable. Her Tanner stage was B1PH3. She did not show café-au-lait spots. **The weight was 52.5 kg (+1.68 SDS), height 161.5 cm (+3.2 SDS, target height -1.01 SDS) and BMI 20.1 kg/m² (+0.64 SDS).**



A **brain CT and MRI** showed a **fibro-osseous lesion of the cranial base**, extending to the orbit and left frontal region with obliteration of the left frontal and sphenoid sinuses and partial obliteration of the ethmoid sinus (Picture above). The pituitary appeared enlarged with a convex upper margin and two small areas of altered signal of the parenchyma with delayed gadolinium uptake (3 and 5 mm in diameter) compatible with pituitary microadenomas and a displacement of the pituitary stalk. The remaining **radiograph of the skeleton** did not show any further lesions.

Endocrine examination showed high IGF-1 and high basal GH levels that resulted not suppressed during OGTT. A hyperprolactinemia was also revealed (Table 1); levels of IGFBP3, TSH, FT4, ACTH, cortisol, blood glucose and electrolytes resulted normal. FSH, LH and 17-beta-estradiol resulted in pre-pubertal ranges. **Pelvic ultrasound** showed a pre-pubertal appearance of uterus and ovaries without ovarian cysts.

	basal values	+45 days	+90 days
IGF-1 ng/ml (123-427)	908	424	599
Basal GH ng/ml (<8.05)	36	11.24	9.96
Nadir GH during OGTT ng/ml (<1)	29.1		10.2
Prolactin ng/ml (4.8-23.3)	451	99	68.6

Table 1. Hormonal values of the patient: basal levels, levels after 45 and 90 days from the start of cabergoline and lanreotide

- ✓ Gigantism
- ✓ GH-PRL secreting adenoma
- ✓ Fibrous dysplasia of bone

Therapy with cabergoline (0.5 mg twice a week) and lanreotide (30 mg every 28 days) was started, with an increase in the dosage after the check at 3 months (cabergoline 0.5 mg 3 times a week, lanreotide 60 mg every 28 days). MRI performed three months after the start of therapy showed a slight reduction in the secreting adenoma. The genetic investigation for McCune-Albright syndrome (MAS) is ongoing.

DISCUSSION

MAS, a complex disorder due to postzygotic somatic activating mutations in GNAS1 gene, is characterized by fibrous dysplasia (98% of patients in a large cohort) (Collins et al,2012), café-au-lait spots (66%) and hyperfunctioning endocrinopathies. The latter are mainly represented by precocious puberty (50%), more rarely by hyperthyroidism (28%), renal phosphate wasting (43%), growth hormone (GH) and/or prolactin hypersecretion (21%) and hypercortisolism (4%) (Yao et al,2017). GH excess represents a serious complication and it is almost always associated with skull base fibrous dysplasia (Salenave et al,2014). Cases of patients treated conservatively are reported in literature (Classen et al,2012).