Clinical Case Reports

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CLINICAL IMAGE

Clitoromegaly as first presentation of a neurocutaneous syndrome in a 3-year-old girl

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Funding Information
No sources of funding were declared for this study.

Received: 26 January 2015; Revised: 16 April 2015; Accepted: 21 May 2015

Clinical Case Reports **2015; 3(9): 767–768**

doi: 10.1002/ccr3.320

Key Clinical Message

A rare cause of clitoral hypertrophy in a child is neurofibromatosis type 1 (NF1). Although evaluation, including karyotype and hormonal studies, is necessary to exclude ambiguous genitalia, the diagnosis of neurofibromatosis as a possible cause of clitoromegaly may help avoid lengthy and sometimes invasive interventions.

Keywords

Child, clitoromegaly, neurofibromatosis type 1.

NF1 is an autosomal progressive disorder having incidence of approximately 1 in 3000 live births. Involvement of the external genitalia in the form of clitoromegaly is extremely unusual [1]. Differential diagnosis consists of hormonal (ambiguous genitalia, precocious puberty, congenital adrenal hyperplasia, masculinizing tumours), nonhormonal conditions (neurocutaneous syndromes, epidermoid cysts, nevus) or could be idiopathic. Current suggested management of a child with clitoral hypertrophy is surgical excision with clitoroplasty at diagnosis to preserve the clitoris and neurovascular bundle. Regular postoperative monitoring for local recurrence is recommended [1]. Renal artery stenosis (RAS) is the most frequent cause of hypertension in NF1 [2]. Children with NF1 should be monitored at yearly intervals. A 3-year -old girl presented with an hypertrophic clitoris noted from birth and growing in size. Coarse facial features, >10 'café au lait' spots (maximum size 4.5 cm × 3 cm) (Fig. 1), enlarged clitoris (4 cm × 1 cm) (Fig. 1), labia normal, head circumference > 97th percentile, mild speech delay, no signs of puberty or hirsutism were noted. Karyotype (46, XX), a short Synacthen test, thyroid function, FSH, LH, oestradiol, testosterone, prolactin, DHEAS and bone age were normal. Magnetic resonance imaging (MRI) of the pelvis and external genitalia revealed normal adrenal glands and female internal genitalia, and a well-circumscribed, pedunculated,

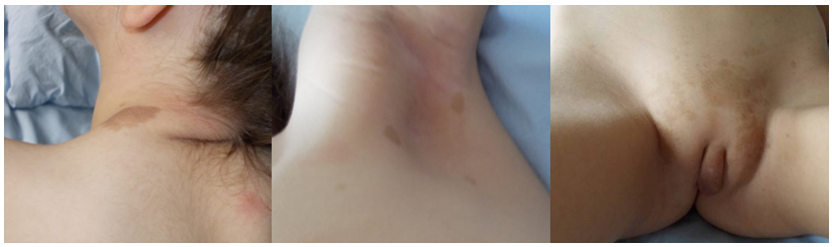

Figure 1. 'Café-au lait' spots and clitoromegaly: signs of NF1 in the child.

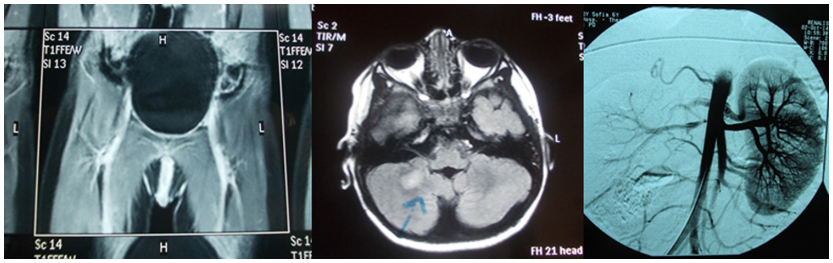

Figure 2. MRI of external genitalia and brain, MRA of renal arteries.

soft tissue mass within the subcutaneous tissues of the clitoral hood (Fig. 2); biopsy confirmed a neurofibromatous lesion. T2-weighted brain MRI showed hyperintense lesions compatible with NF1 (Fig. 2). In due course, the patient developed renovascular hypertension due to right renal artery stenosis (Fig. 2, Magnetic Resonance Angiogram (MRA)). She was managed by a multidisciplinary team of pediatric subspecialties and neurofibroma excision with clitoroplasty was performed.

Conflict of Interest

None declared.

References

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