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Pediatrics

Clitoromegaly in a Pediatric Female Patient Associated with Congenital Adrenal Hyperplasia: A Case Report

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Abstract **Case Report**

Background: Congenital adrenal hyperplasia (CAH) is most common cause of virilization and clitoromegaly in genetically female children. Excess androgen production during fetal life may result in enlargement of the clitoris, sometimes resembling a penile structure. CAH due to 21-hydroxylase deficiency accounts for most cases of genital ambiguity in 46 females. [1,2] We report a case of 3-year-old female child, previously diagnosed with congenital adrenal hyperplasia, presented with clitoromegaly. Clinically, the enlarged clitoris resembled a small penis. Surgical reduction clitoroplasty was performed. Histopathological examination of the excised specimen showed benign hypertrophy of clitoral tissue. No evidence of malignancy, dysplasia, or neoplastic lesion was identified. ***Conclusion:*** Clitoromegaly in children is most commonly associated with congenital adrenal hyperplasia. Histopathological examination confirms the diagnosis by demonstrating normal clitoral erectile tissue with hypertrophy secondary to androgen exposure. Early diagnosis and multidisciplinary management are important for optimal functional and cosmetic outcomes.

Keywords: Congenital adrenal hyperplasia, clitoromegaly, virilization, Pediatric pathology, histopathology, clitoroplasty.

INTRODUCTION

Congenital adrenal hyperplasia (CAH) is autosomal recessive disorders caused by defect in adrenal steroidogenesis. Deficiency of 21-hydroxylase is mainly responsible for approximately 90–95% of cases. Impaired cortisol synthesis leads to increased adrenocorticotropic hormone (ACTH) secretion and adrenal hyperplasia, resulting in excess androgen production. [1]

Female children with classic CAH may present with ambiguous genitalia, labial fusion, and clitoromegaly. The severity of virilization varies from mild enlargement of the clitoris to a penile-like phallus. Histopathological descriptions of excised clitoral tissue are relatively uncommon in the literature, making such reports valuable for pathologists and clinicians. [2,3]

CASE REPORT

A 3-year-old female child visited to the pediatric surgery department with enlargement of the external genitalia since 4 months. The patient was a known case of congenital adrenal hyperplasia.

Physical examination revealed a markedly enlarged clitoris measuring approximately 4 cm in length, resembling a small penis. No palpable gonads were identified. The external genitalia otherwise showed female characteristics. Reduction clitoroplasty was done. Excised specimen was sent for histopathological examination.

In the pathology department, we received a skin covered soft tissue mass measuring 4 × 2 × 2 cm. Cut section shows grey brown to grey tan areas (Fig 1 and Fig 2).

Figure 1

Figure 2

On Microscopic examination- Sections studied shows keratinized stratified squamous lining epithelium (Fig 3.) The subepithelium is composed of fibromuscular and fibroconnective tissue along with presence of numerous cavernous blood vessels showing benign hypertrophy of erectile tissue which was consistent with clinical diagnosis of Clitoromegaly (Penile like structure)"

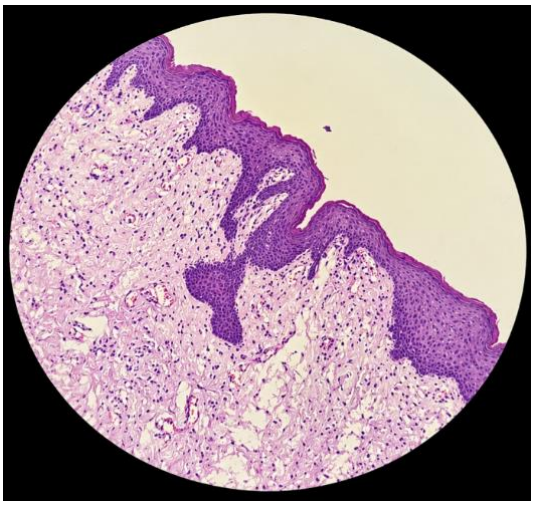

Figure 3: H&E (200x) shows keratinized stratified squamous lining epithelium

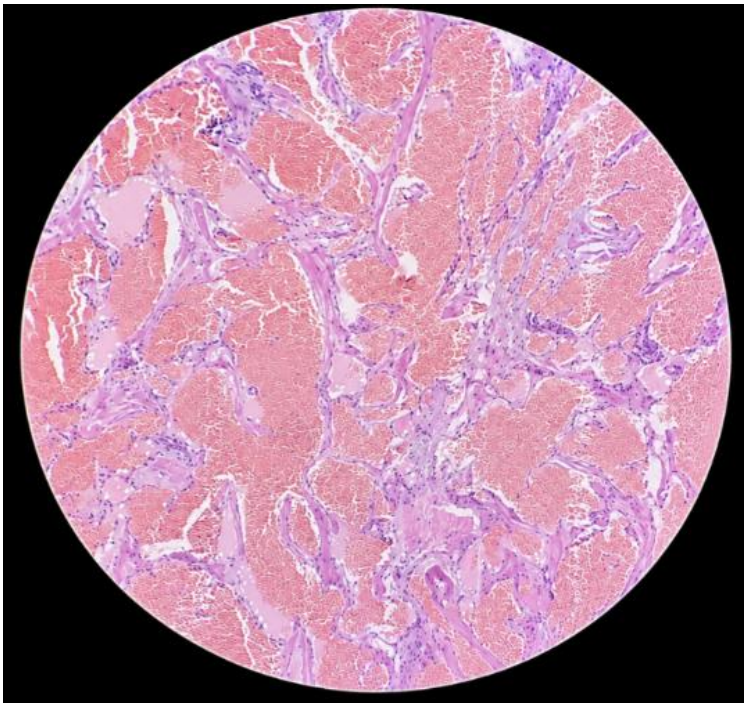

Figure 4: H&E (200x) shows numerous cavernous blood vessels

DISCUSSION

Clitoromegaly is an abnormal enlargement of the clitoris and is usually caused by excess androgen exposure during fetal life, infancy, or childhood. Congenital adrenal hyperplasia remains the most common underlying cause in pediatric patients. [1,4]

In the present case, a 3-year-old female child with known CAH developed marked clitoromegaly that clinically resembled a penile structure. Histopathological examination demonstrated hypertrophied erectile tissue and vascular sinusoids without any neoplastic change.

Several authors have reported similar findings in young children. A recent case reported by Sharma *et al.,* described a young girl with CAH-associated clitoromegaly who underwent clitoroplasty. The authors emphasized that excessive prenatal androgen exposure was responsible for genital virilization and enlargement of the clitoris. [5]

Heng and Eugster et al reported a 2-year-old girl presenting with rapid growth and clitoromegaly due to androgen excess. Their report highlighted the importance of early endocrine evaluation in toddlers presenting with genital enlargement. [6]

A 2025 newborn case reported by Bhatia *et al.,* described apparent clitoromegaly at birth that led to the diagnosis of congenital adrenal hyperplasia. Early recognition allowed prompt hormonal treatment and prevention of adrenal crisis. [7]. Boccardo and colleagues reviewed clitoromegaly in childhood and adolescence and noted that androgen excess during fetal life is the principal mechanism responsible for clitoral enlargement. They stressed the importance of distinguishing hormonal causes from tumors and other rare conditions. [4]

In older females, long-term studies have shown that clitoromegaly may persist despite hormonal therapy. Szymanski *et al.,* evaluated women with CAH and found that clitoromegaly significantly affected body image and quality of life, emphasizing the importance of individualized management. [8]

Histologically, clitoromegaly associated with CAH typically shows normal clitoral architecture with enlargement of erectile tissue, vascular spaces, and smooth muscle elements. The absence of atypia or neoplastic change supports a non-neoplastic androgen-mediated process. These findings were also observed in our patient.

The present case highlights the characteristic histopathological features of clitoromegaly in a young child with CAH and contributes to the limited literature on tissue-based evaluation of this condition.

CONCLUSION

Clitoromegaly is a common manifestation of congenital adrenal hyperplasia in genetically female children. Severe enlargement may resemble a penile structure and create diagnostic and psychosocial challenges. Histopathological examination typically reveals hypertrophied erectile tissue with preserved architecture and no evidence of malignancy. Early diagnosis, endocrine treatment, and multidisciplinary management are essential for favourable outcomes.

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