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RESEARCH ARTICLE

HYPERTROPHY OF THE CLITORIS IN AN ADULT WOMAN (CASE REPORT)

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Abstract

Non-classical congenital adrenal hyperplasia by 21 hydroxylase deficiency is an autosomal recessive disease whose usual presentation is a late virilization. In some African countries like Morocco, there are cultural barriers to gynecological examination in girls, women consult until late for a sexual problem as in the case of our patient who consulted for a clitoris hypertrophy at the age of 22 years and the hormonal assessment found a high level of 17 hydroxerone in favor of a 21 hydroxylase deficiency.

Introduction:-

Non-classical congenital adrenal hyperplasia causes excessive levels of androgens in women and may manifest as hirsutism or verilization of the external genitalia.

In some African countries like Morocco, there are cultural barriers to gynecological examination in girls, women consult late for a sexual problem as in the case of our patient who consulted for clitoris hypertrophy and primary amenorrhea

Observation:-

A 22 year old female patient, from a first degree cosanguineous marriage, consulted for hirsutism associated with primary amenorrhea

The examination found: a short stature 150cm (-2DS), an excessive growth of terminal hair in a man-like pattern as a hirsutism rated at 27 (according to the Ferriman and Galoway score) especially on the chin (image 1). Signs of virilization: Male morphotype (mammary atrophy, increase of muscle mass) and ambiguous external genitalia: clitoris hypertrophy of 2cm, urethral orifice in median position with presence of vaginal orifice in normal position, no palpation of testis at inguinal level (image2).

Hormonal assessment: Total testosterone elevated to 2.48 ng/ml (0.14 - 0.68), DHEA dehydroepiandrosterone sulfate elevated to 7073 ng/ml (1480-4070) and 17 Hydroxy-progesterone elevated to 58.8ng/ml which confirmed the diagnosis of non-classical congenital adrenal hyperplasia due to 21-hydroxylase deficiency

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CT scan showed normal sized and homogeneous adrenals without mass or hyperplasia. Pelvic MRI showed: female internal genitalia, uterus of normal size (61mm) and ovaries of normal size.

The patient was put on treatment to slow down adrenal androgens with hydrocortisone and was referred to gynaecology for reconstructive surgery of the clitoris hypertrophy.

Discussion:-

Non-classical congenital adrenal hyperplasia affects 1-10% of hyper androgenic women depending on ethnic and geographic origin (1) . It is secondary in 90-95% of cases to 21-hydroxylase deficiency related to mutations in the CYP21A2 gene with autosomal recessive transmission (2).

The 21-hydroxylase block in the "non-classical form" is a partial enzymatic deficiency in 21-hydroxylase leading to an excess of androgen precursors, 17-hydroxy-progesterone (17OHP), partly converted into adrenal androgens responsible for hyperandrogenism (3) as in the case of our patient who had an elevated level of 17OHP responsible for the signs of virilization.

At the hypothalamic level, 17-hydroxy-progesterone and androgens converted to estrogens exert a negative feedback control on the gonadotropic axis causing primary amenorrhea in our patient.

The non-classical form of 21-hydroxylase block can manifest itself in adolescence by hirsutism, which generally appears in the peripubertal period and is the most frequent sign, acne, alopecia, an android morphotype (increase of muscle mass in a rather short woman , excessive growth of terminal hair in a man-like pattern). A masculinization of the external genitalia can sometimes be seen in the form of clitoris hypertrophy [4] as in the case of our patient.

Pelvic imaging (ultrasound or pelvic MRI) confirms the presence of the female genital organs and ultrasound can find a sonographic appearance of micropolycystic ovaries secondary to hyperandrogenism, some of the excess adrenal androgens act at the ovarian level and would be able to induce a disorder of folliculogenesis, similar to that observed in micropolycystic ovary syndrome, by acting on the granulosa cells of the growing ovarian follicles. An ultrasound picture of polycystic ovaries has been reported in up to 50% of patients with a non-classical form of 21-hydroxylase block (therefore 21-hydroxylase block should always be ruled out before making the diagnosis of micropolycystic ovary syndrome, in accordance with the Rotterdam criteria), which should not be confused with micropolycystic ovary syndrome which is a diagnosis of elimination (5).

Biological diagnosis is based on a baseline 17hydroxy progesterone level greater than 2 ng/ml or, if normal, a concentration > 10 ng/ml after the synacthen test [2,8]. The data concerning the appearance of the adrenals in the late form remain very specific and are in favour of hyperplasia with or without a nodular component. In our patient, the adrenals were normal on CT scan (6).

Treatment is based on a moderate dose of hydrocortisone (20 to 30 mg per day) or dexamethasone (0.5 to 1 mg per day) which slows down the secretion of ACTH (Adrenocorticotropic Hormone) and therefore slows down adrenal androgens.

He use of anti-androgens such as cyproterone acetate is proposed in cases of disabling hirsutism associated with cosmetic treatment by laser hair removal (7). In the case of external genitalia anomaly, reduction of clitoral hypertrophy while preserving sensitivity should be done early.

If the patient is planning a pregnancy, it is essential to know the genetic status of the partner, since 21-hydroxylase deficiency is an autosomal recessive disease and the frequency of heterozygous (and therefore healthy) patients is 1/50 in the general population. If a mutation on one of the two alleles is detected in the partner, the analysis of the genotype of both parents makes it possible to evaluate the risk of transmission of a severe form and to envisage adequate antenatal and neonatal management if the foetus is affected. The objectives of this management are to prevent the risk of virilization of a female foetus and to anticipate the risk of Salt-wasting Syndrome of the Newborn [8].

Conclusion:-

Late-onset congenital adrenal hyperplasia is one of the diagnoses that should be routinely sought in women who present with signs of hyper androgenism with virilization. The diagnosis should be made as early as possible to allow normal growth, female puberty and satisfactory fertility. Therefore, it is important to educate young girls to consult in case of any sexual problem.

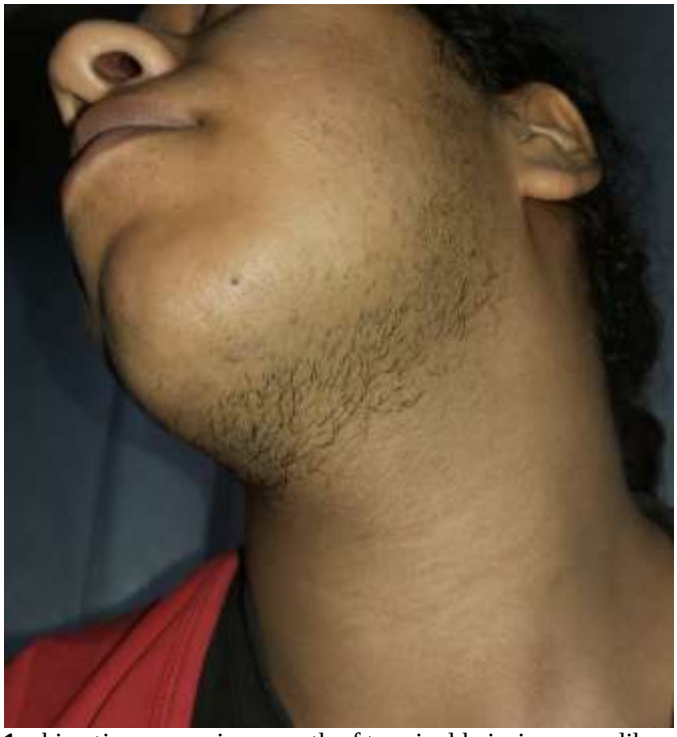

Figure1:- hirsutism excessive growth of terminal hair in a man-like pattern.

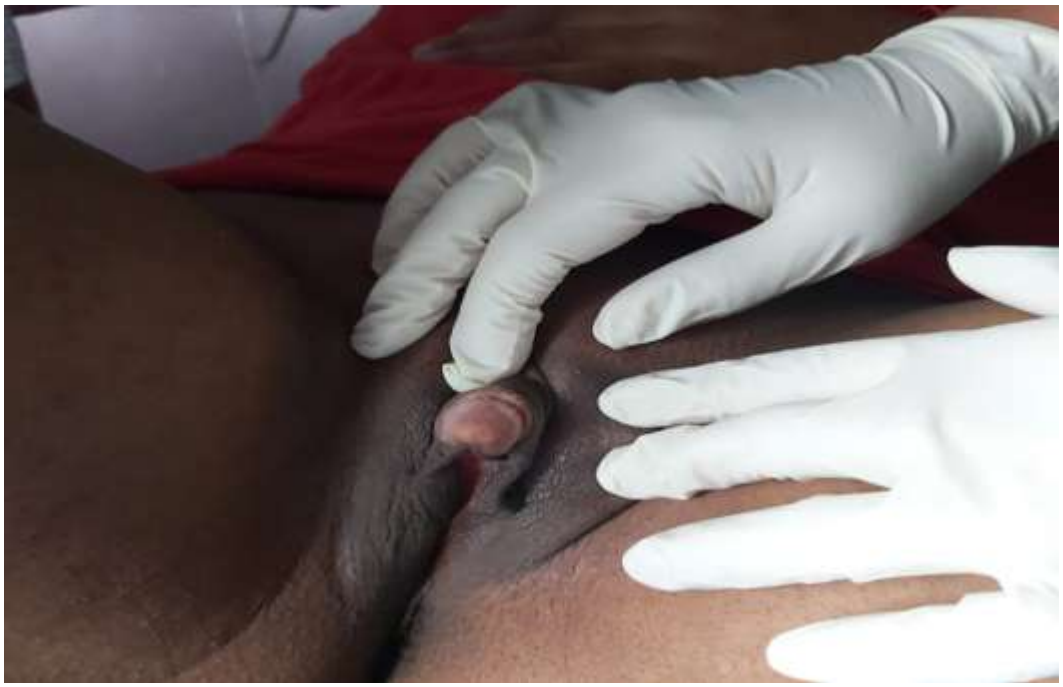

Figure 2:- Clitoris hypetrophy.

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All authors conceptualized and wrote the paper, revised the text; and all approved the final manuscript.

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