ESPE 2021
The 59th Annual Meeting Online
22-26 September 2021

P2-419 A LATE REVELATION OF SEXUAL DIFFERENTIATION DISORDER 46XX DUE TO 11 BETA HYDROXYLASE DEFICIENCY

SABRINA KHENSAL, AMINA BOUHLASSA, CHAHINEZ HABI, NASSIM NOURI

Endocrinology Department, Constantine, Algeria

UNIVERSITE DE CONSTANTINE 3
جامعة قسنطينة 3

INTRODUCTION

Congenital adrenal hyperplasia due to 11 beta hydroxylase deficiency, is an autosomal recessive pathology that represents 5 to 8% of congenital adrenal hyperplasia.

The predominant symptoms of this disorder are androgen excess' features, including masculinization of female newborns and precocious puberty in male children. Approximately, two thirds of patients also have hypertension, which may or may not be associated with mineralocorticoid excess, hypokalemia, and metabolic alkalosis.

This observation reports the case of a child raised as a male, with congenital adrenal hyperplasia due to 11 beta hydroxylase deficiency, discovered as part of an exploration of a precocious pseudo puberty with high blood pressure and hypokalemia.

OBSERVATION

B.M is a 9 years old patient, from a second-degree consanguineous marriage, with an assigned male sex. B.M was brought up as a boy, he had a personal medical history of rheumatic fever and a similar case with his German cousin has been described. His height was 136cm (-1DS), Tanner was A5P5S1 with bilateral unpalpable testes.

BP: 170 / 120 mmHg. Karyotype test result was 46 XX, and laboratory examination showed: a high value for ACTH: 374 pg/ml, testosterone: 4,3ng/ml delta 4-androstenedione: 126,4 nmol/l and Deoxycorticosterone (DOC): 1448 pg/ml. Ovaries and a uterus fundus, without abnormalities were found on USG examination. CT showed a homogeneous bilateral adrenal Hyperplasia. Transthoracic ultrasound: concentric left ventricular hypertrophy, minimal mitral regurgitation, minimal aortic insufficiency. Bone age study corresponded 17-year-old male. The primary psychiatric expertise affirmed that the child is oriented towards the male phenotype and her parents chose to keep him as a male.

The orientation of child's sex will be reassessed after one year of psychiatric follow up. Removal of internal genitalia and wide hysterectomy before puberty will be, therefore, discussed.

DISCUSSION

Late diagnosis of Congenital adrenal hyperplasia is a problem of sexual identification, this is why diagnosis and antenatal treatment are essential to avoid the virilization of the feminin sex fetus. The identification of the absence of testicles in the apparent scrotum and inguinal canal at birth, could have led to an early diagnosis and avoided dealing with a mismatch between the chromosomal and the assigned sex at a late age, which is a real dilemma in this case.

The challenge is to ensure good growth, to reduce androgen hyperstimulation by corticoid therapy and to ensure a good quality of life.

REFERENCES

1 - Houk CP, Lee PA: Consensus statement on terminology and management: disorders of sex development. Sex Dev 2008;2:172–180.

2- Lee PA, Houk CP, Ahmed SF, Hughes IA: Consensus statement on management of intersex disorders. International Consensus Conference on Intersex. Pediatrics 2006;118:E488–E500.

3- Lin-Su K, Lekarev O, Poppas DP, Vogiatzi MG: Congenital adrenal hyperplasia patient perception of 'disorders of sex development' nomenclature. Int J Pediatr Endocrinol 2015;2015:9.

CONTACT INFORMATION

skhensal51@yahoo.fr.