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Case Report

Van Wyk Grumbach syndrome: a case report

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ABSTRACT

Van Wyk Grumbach Syndrome is characterized by long standing hypothyroidism, precocious puberty, Ovarian cysts and delayed bona age. We present a case of a 12 year old female who presented with Short stature and precocious puberty. The Patient responded well to L- thyroxine therapy.

Keywords: Van Wyk Grumbach syndrome, Hypothyroidism, Precocious puberty

INTRODUCTION

Van Wyk Grumbach syndrome is characterized by juvenile hypothyroidism, delayed bone age and isosexual precocious puberty with reversal to pre pubertal state following thyroxine replacement therapy.[1] We report a case of 12 year old girl who presented to our OPD with short stature, constipation and precocious puberty due to long standing hypothyroidism.

CASE REPORT

A girl aged 12 years presented to our OPD three months back with complains of not gaining height adequately, constipation and poor scholastic performance. She had attended menarche at the age of 8 years, the menstrual cycles were irregular and the flow was scanty. She was born out of a normal vaginal delivery and was the first of two siblings. Her parents were of normal height and weight. There was no history of headaches, visual disturbances, vomiting and head trauma.

Her height was 88.5 cm (expected median height being 151 cm), Upper segment to lower segment ratio 1.3:1, weight was 20.4 kg. Pulse rate was 62/min; Blood pressure was 100/74 mm Hg. On examination she had pallor, coarse facial features, large tongue, facial hirsuitism, dry skin. According to Tanner staging her Sexual maturation score was B4, P3. Visual acuity and fundus examination was normal. Her investigation were as follows Hb 7.4 gm/dl, MCV 97 fl, RBS 79 mg/dl, blood Urea 18 mg/dl, serum creatinine 0.75 mg/dl, T_3 0.324 ng/ml (normal 0.7-2.0 ng/ml), T_4 0.462 mcg/dl (normal 5.5-13 mcg/dl), TSH >100 mic IU/ml(Normal 0.27- 4.20 micIU/ml), LH 2.16 mIU/ml, FSH 6.6 mIU/ml, Serum Prolactin 100.2 ng/ml (2.8-28). Her radiological investigations revealed a bone age of two years. Ultrasound of the pelvis showed a uterine size of 7.29×2.55×5.10 cm. Uterus was anteverted, normal in size, shape and echo density. Both the ovaries were enlarged and multiple cystic septate areas. MRI of the patient could not be done due to financial constraints. The Patient was started on thyroxine replacement therapy and was found to have improvement in symptoms when reviewed after three months.

DISCUSSION

Sexual precocity is associated with an increase in linear height, premature epiphyseal fusion ultimately leading to short stature. Von wyk and Grumbach in 1960 first described the association of long standing hypothyroidism , isosexual precocious puberty and poly cystic ovaries.[1,2] The precocious puberty seen in this syndrome is always isosexual and incomplete. In girls the

condition usually presents with irregular menstrual bleed and uncommonly with breast enlargement and galactorrhoea.[3] Pubic hair are conspicuous by their absence. In boys usually it is macroorchidism without presence of axillary hair. Early recognition of this entity is important as the early institution of thyroid hormone replacement therapy can help in resolution of symptoms. The most common cause of hypothyroidism in these patients is auto immune thyroiditis. Von wyk grumbach syndrome can be diagnosed by its salient clinical and radiological features.

Figure 1: Van Wyk Grumbach syndrome.

The exact mechanism as to why precocious puberty develops in von wyk grumbach syndrome is speculative. Postulated mechanism is lack of specificity in feedback mechanism leading to overproduction of multiple hormones. TSH levels are consistently elevated in such patients and the manifestations of sexual precocity might be directly related to TSH levels. Low FSH like activities of TSH becomes significant at high concentrations of TSH. Hyperprolactinemia, reduced gonadotropin clearance and decreased dopaminergic and opioid tone at the hypothalamic pituitary axis are other plausible explanations of precocious puberty. The increased FSH or FSH like activity of TSH causes a high FSH/LH ratio which is in contrast to high LH/FSH ratio which is there in normal puberty, the increased FSH/LH ratio causes increased estrogen secretion thereby leading to multiple follicles in the ovaries. Direct effect of Thyroxine on prepubertal testis, which leads to proliferation of sertoli cells, is responsible for macroorchidism in males. The common causes of incomplete sexual precocity are intake of hormonal preparations, sex hormone secreting tumors, McCune Albright Syndrome. However in all above cases the bone age is advanced. Delayed bone age along with hypothyroidism suggests a strong possibility of Van wyk grumbach syndrome.

Rastogi A et al, have reported a case of 10.7 year old female suspected of having van wyk grumbach syndrome having responded successfully to thyroxine therapy.[4] Branoswki et al have reported a 8 year old girl with long standing hypothyroidism who presented with features suggestive of von wyk grumbach syndrome.[5] Sneha LM et al, have reported a case of short stature diagnosed as having van wyk grumbach syndrome which responded well to thyroxine therapy.[6]

Although the etiology of the disease remains unclear the treatment plan remains the same with patients responding well to thyroxine therapy. Patients respond well to thyroxine therapy with improvement in symptoms and resolution of symptoms.[7] We report this interesting association between hypothyroidism and precocious puberty as it would help fellow colleagues in reaching to a diagnosis sooner. The case is being reported for its rarity.

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