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PRIMARY HYPOTHYROIDISM PRESENTING AS SHORT STATURE AND DELAYED MENARCHE

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INTRODUCTION

Short stature, is defined as height or length below 2.5th centile or less than 2 standard deviation for that specific age and sex.[1,2] Short stature is defined as an adult height of 4 feet 10 inches or under, as a result of a medical or genetic condition. Although other groups may extend the criteria for certain forms of short stature to 5 feet, the average height of an adult with short stature is 4 feet.

Growth is affected by genetic, environmental, systemic and endocrine influences. Medical care depends up on the aetiology of the short stature. Primary hypothyroidism is one of the endocrine factors responsible for short stature.[3] Primary hypothyroidism constitutes one of the major treatable causes of short stature, which frequently goes unrecognized and not treated in some parts of the world. Hypothyroidism constitutes about 8-10% cases of short stature in the developed nations[4], while it is a major problem in developing part of world where it constitutes 25-30% cases of short stature.[5]

Hypothyroidism is a deficiency in thyroid hormone secretion by the thyroid gland, with a reduction in thyroid hormone action at the cellular level. The two major forms in children are: 1) congenital primary hypothyroidism (occurring in 1:1700 live births) and 2) acquired hypothyroidism due to diseases that have an onset any time after birth. Serum TSH concentration and free T4 are the most sensitive screening tests for primary hypothyroidism.

Common endocrine disorders leading to short stature include; hypothyroidism, Cushing's syndrome and growth hormone deficiency.[6]

KEYWORDS: Primary hypothyroidism, short stature, delayed menarche, mental retardation.

CASE REPORT

A patient named XYZ was admitted to AVBRH, JNMC Sawangi which is a rural medical college with the complaints of pain in abdomen and excessive menstrual bleeding. This was her first menses. She also had epigastric pain and two episodes of vomiting. She had chest pain which was not radiating. There was no history of shortness of breath or palpitations. The patient does not have complaints of any major illness in the past. There is no history of short stature in the family. Her parents and brothers were of normal height and development. There was history of delay in developmental milestones. Walking and speaking started at age of four years of age. Later she went to school up to third standard and then dropped out from school.

She was an 18 year old girl who was short in stature, height 96 cm and weight 15 kg. She had normal facial features, normal trunk and proportionate hands and limbs. Secondary sexual characters were well developed. Breast development was normal and pubic and axillary hair were present. She had slow speech and dry skin. She was examined by a clinical psychologist and IQ testing was done which gave IQ of 31 which indicates severe mental retardation. Ophthalmic examination was normal.

On laboratory investigations – the haemoglobin was found to be 3.4 gm%, HCT was 11.1, TLC was 8100, platelets were 1.72 lakhs/cmm. Blood sugar levels, kidney function tests and liver function tests were normal. X-ray chest showed cardiomegaly with left ventricular hypertrophy. ECG was within normal limits. 2D-ECHO was done which showed mild to moderate pericardial effusion, there was no evidence of cardiac tamponade and function of both the ventricles was normal. On ultrasound the uterus was 8×2.5×2.1 cm, endometrial thickness was 4mm both the ovaries were normal. Other abdominal structures the liver, spleen, and both kidneys were of normal size and shape. X-ray of spine, skull and upper and lower limbs was done which did not show any abnormality. There was osteoporosis and bone scan indicated delayed bone age. Her growth hormone was within normal limits. TSH was >150, T3 was < 0.30, T4 was 0.90.

In view of anaemia 75 ml of compatible blood was transfused. Haemoglobin was repeated after 2 days which was 8.4 gm%. Eltroxin was started in the dose of 50 mg once daily. The plan was to gradually increase the dose till the TSH levels drop. She was also advised supplementation with iron, calcium and vitamin D. Patient is being followed up regularly.

DISCUSSION

Short stature can be caused by more than 200 conditions. It can also be due to familial short stature, constitutional delay of growth and development, glucocorticoid therapy, gastro-intestinal and renal diseases, immunological factors and certain cancers. Causes of proportionate short stature include metabolic and hormonal disorders. Most occurrences of short stature result from a random genetic mutation in either the father's sperm or the mother's egg rather than from either parent's complete genetic makeup. Genetic diseases did not seem likely in this patient as she did not have facial dymorphology, there was no thoracic deformity nor disproportionate limbs. The patients close relatives, her mother, father and brothers were of normal stature. Also she did not have any other major organ pathology.

Because the TSH levels were very high in our patient, the diagnosis of primary hyperthyroidism was made. Hypothyroidism is caused by a deficiency in the synthesis of thyroid hormone. Short stature is the most obvious skeletal manifestation. When the skeleton is affected, the severity of this manifestation depends on the degree of the deficiency and age of onset.

Treatment of hypothyroidism at least five years before the onset of puberty is essential to attain a height consistent with normal. Our patient presented to us very late. However she was proportionately developed although of short stature. Also her gynaecological examination was normal.

CONCLUSION

Primary hyperthyroidism is an important aetiology for short stature, delayed puberty and mental retardation. If diagnosed early before the onset of puberty, supplementation with thyroxin will lead to normal physical and mental development.

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