Case Report

Congenital Hypothyroidism Presenting as Menorrhagia in Adulthood

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Abstract

Although hypothyroidism is a common cause of menorrhagia, it is an uncommon presentation of congenital hypothyroidism. We report a case of congenital hypothyroidism presenting in adulthood with severe menorrhagia. Despite the late presentation, she had features to suggest hypothyroidism since birth.

Introduction

Congenital hypothyroidism (CH) is a frequently occurring condition with varied onset and clinical features. It has a worldwide incidence of 1:4000 live births with estimated incidence in India being 1:2500-2800 live births. Most of the cases are due to thyroid ectopia, aplasia or hypoplasia and are sporadic in occurrence with a 2:1 female to male preponderance.

History

A 19 year old female belonging to Muslim community from the union territory of Puducherry presented to us with history of menorrhagia for 1 month prior to admission. She was the second child of a non-consanguineous marriage. Her developmental milestones were delayed since birth although her delivery was normal and at term. She was able to sit unaided only at 18 months and walk only at 3 years. At around the same time, mother noticed that she had developed a protuberant abdomen, puffiness of face and dry skin. Her level of scholastic performance was poor; she could not read or write. She could only manage simple household work. Her growth remained stunted, and her menarche was delayed until the age of 19 years. There was no history of thyroid dysfunction or mental retardation in her family.

On examination her vitals were stable, she had stunted growth with height of 98 cm (< 3rd percentile), weight of 25 kg (< 3rd percentile) and her limbs were short with upper segment to lower segment ratio of 57/42. She had typical features of hypothyroidism (Figure 1a) with facial and periorbital puffiness, cold peripheries, coarse thick dry skin, ankle and biceps tendon jerks showing delayed relaxation. Thyroid gland was palpable in the neck moving with deglutition. Her sexual maturity rating (SMR) was breast development at Tanner stage 4 and pubic hair at Tanner stage 3. Abdomen and pelvic examination was normal. Her mental age as assessed by Binet - Kamat Test (BKT) of Intelligence was 3 years 8 months which approximated her height age (3 years 9 months).

Investigations revealed the following findings: she had severe anaemia with haemoglobin of 5.1 gm/dl (normal 12.0–15.8 gm/dl) with peripheral smear showing microcytic hypochromic RBCs and low serum ferritin of 12 ng/ml (normal 50-200 µg/L). Her thyroid profile was suggestive of primary hypothyroidism with free T₄ - 0.4 pg/ml (normal 2.3 – 4.2 pg/ml), free T₃ - 0.10 ng/dl (normal 0.89 – 1.76 ng/dl), and TSH >150 µIU/ml (normal 0.35 – 5.5 uIU/ml). Total cholesterol was 346 mg/dl (normal <200 mg/dl). Her liver function, renal function, bleeding and clotting time was normal. Ultrasonography revealed the presence of a small thyroid gland (each lobe: 1x2 cm) with altered coarse echoes and 99m-Technicium pertechnate thyroid scintiscan showed no uptake in normal thyroid bed with normal salivary gland and background activity (Figure 2). Her bone age assessed radiologically by Greulich-Pyle method was 12 years suggestive of delayed skeletal maturity. Her Chest X-ray, echocardiography and audiometry were normal.

This 19 year old female was brought to the hospital for the first time. In view of the clinical features with hypoplastic thyroid gland in the background of nil uptake on thyroid scintiscan a diagnosis of congenital hypothyroidism secondary to thyroid dysgenesis was made. The diagnosis of iodine transport defect was unlikely in our patient in view of normal tracer uptake by salivary glands. However the rare possibility of TSH receptor defect could not be ruled out. Her severe anaemia was secondary to blood loss due to menorrhagia. She was managed with transfusion with two units of packed RBCs and haematinics. In view of her long standing hypothyroidism she was started on low dose thyroxine with 25 mcg and gradually increased to 100mcg per day. Her menorrhagia stopped and patient (Figure 1b) is doing well with normalisation of thyroid profile and menstrual cycles.

Discussion

Menorrhagia, as defined by blood loss of 80 mL or more per cycle, affects 9% to 14% of women and has multiple causes. Among the endocrine causes, thyroid disease merits special mention. Though menorrhagia is the most common menstrual
irregularity in hypothyroid women, it is uncommon as a presentation of congenital hypothyroidism in adulthood.

Primary hypothyroidism is characterised by elevated thyroid stimulating hormonal (TSH) levels with normal gonadotropins. TSH has both follicle-stimulating hormone (FSH) and luteinising hormone (LH)–like effects through their shared α-subunit. The LH receptor is stimulated due to “spillover” effect of elevated TSH. As a result, negative feedback is down-regulated, leading to a decreased secretion of luteinising hormone resulting in decrease in progesterone. In addition, sex hormone binding globulin levels are low which in turn leads to elevated levels of circulating free oestrogens. This prolonged, unopposed hormonal effect on the endometrium results in failure of ovulation and corpus luteum formation, ultimately leading to menorrhagia. Other factors that may contribute to excess bleeding are defective intrinsic clotting mechanism due to decreased concentrations in plasma of factors VIII and IX, acquired von Willebrand’s disease, increase in capillary fragility and the decrease in platelet adhesiveness. Notably, both symptoms and laboratory aberrations tend to normalise after treatment with thyroxine, as in our case menorrhagia stopped after normalisation of thyroid hormone levels.

Congenital hypothyroidism is a common preventable cause of mental retardation. Early diagnosis of congenital hypothyroidism by mass neonatal screening programmes is of the foremost importance for the prevention of long-term sequelae. Our patient had severe mental retardation with growth failure, along with classic features of hypothyroidism like puffiness of face, hoarse voice, dry skin etc. Our patient is unusual in that despite the evidence of hypothyroidism at birth and during childhood the diagnosis was delayed until she developed menorrhagia which led to the detection of congenital hypothyroidism at the late age of 19 years. In an era where we are moving towards universal neonatal screening for congenital hypothyroidism, it is unfortunate to diagnose CH in adulthood only following menorrhagia having missed the glaring diagnosis since birth. It is hoped that more stress can also be laid on public awareness of the conditions like CH so that they are recognised and treated early.

References