International Journal of Basic, Applied and Innovative Research
IJBAIR, 2015, 4(1): 15 - 19
ISSN: 2315 - 5388 E-ISSN: 2384 - 681X
www.arpjournals.com; www.antrescentpub.com

CASE REPORT

CONGENITAL HYPOTHYROIDISM IN A FIVE YEAR OLD NIGERIAN GIRL: A CASE REPORT,

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Received: 30th January, 2015 **Accepted: 27th March, 2015** **Published: 31st March, 2015**

***Endorsed By**: Innovative Science Research Foundation (ISREF). **Indexed By**: African Journal Online (AJOL); Texila American University; Genamics; Scholarsteer; EIJASR; CAS-American Chemical Society*; and *IRMS Informatics India* (*J-Gate)*

ABSTRACT

Congenital hypothyroidism (CH) is a condition of thyroid hormone deficiency present at birth, with an incidence of 1: 3000 - 4000 infants worldwide. It usually manifest in neonatal period or early infancy. The diagnosis can be delayed where routine screening is not done. Untreated infants have profound mental retardation and severe growth restriction. This case report presents a 5 year old girl seen with poor growth, speech and learning difficulties. She was the smallest in class and cuddled by her mates. Her speech was muffled and could not read nor write like her mates. She had delayed milestones, poor growth and reduced activity compared to siblings. Examination revealed anthropometric measurements below the third percentile. She had macroglossia and coarse dry skin. TSH was elevated (>50(µi.u/ml), while T3 and T4 were both unrecordable. Thyroid ultrasound scan revealed hypoplasia, while x-rays and echocardiography showed delayed bone age and mild pericardial effusion respectively. L-thyroxine therapy was commenced, and her growth velocity, school performance and TFT profile over six years improved remarkably. We advocate for routine newborn screening in developing countries including Nigeria, considering the severity of growth stunting and severe mental retardation in CH when diagnosis is missed or treatment delayed.

Key Words: Congenital hypothyroidism, Thyroid hormone, Mental retardation, Growth restriction

INTRODUCTION

Congenital hypothyroidism (CH) is a condition of significant decrease in or absence of thyroid function present at birth (Gomella *et al* 2013). It has a worldwide incidence of 1:3000 - 4000 infants, with a female preponderance of 2:1 (Le-Franchi, 2011) Physiologically, thyroid hormone is critical for normal cerebral development in the early postnatal months; implying that in CH, biochemical diagnosis must be made soon after birth, and effective treatment initiated promptly to prevent irreversible brain damage (UK Newborn Screening 2005; Le-Franchi, 2011).

Available literature indicates that CH is the commonest endocrine and most treatable cause of mental retardation. It also manifests with severe growth retardation. Associated deficits include hearing and visual impairment. It is usually asymptomatic at birth but detectable by newborn screening. Most affected children develop clinical signs during the first few weeks or latter part of infancy, depending on the cause and severity of the deficiency. On rare occasions, symptoms may appear in the second year of life or later (Le-Franchi, 2011; Kayode-Adedeji, 2015).

Generally, CH is under-diagnosed in developing countries including Nigeria, due to absence of screening programmes, poor access to and high cost of laboratory diagnostic investigations. In developing countries, the index of suspicion for this condition is low in the evaluation of children with failure to thrive or mental retardation. Good outcome is hinged on early diagnosis and prompt treatment in the first few months of life.

The case being presented is that of a girl delivered to parents of high socio-economic status who appeared to be asymptomatic till about two years of age; presented at five years of age and had her treatment monitored for six years. We further seek to draw attention to this disease and advocate for a screening programme in developing countries, Nigeria inclusive.

CASE PRESENTATION

A five year old girl presented to our institution with complaints of poor growth, speech and school performance of about three years duration. Parents noticed that she had not been growing for about three years; she had the same height as her 20 month old sibling and was the smallest in her class. She had muffled speech and could only muster two to three word phrases. She was described as slow in learning and could neither read nor write like her classmates.

She had been noticed to have poor appetite in the same period and passed stools which were sometimes hard, two to three times weekly. She had cold intolerance and was not as active as her siblings. She was delivered at term with a birth weight of 2700gm and did not have prolonged jaundice, poor feeding or lethargy. Her milestones, though at the lower limit of normal, lagged behind her siblings. Her nutrition in infancy was adequate. Her mother lived in the goitre belt but did not have neck swelling nor used anti-thyroid medication. Her siblings did not have similar problems.

Clinical examination revealed a girl who was small for age with short neck, macroglossia, and cool, dry, rough and thick skin. Her weight, height and BMI were 10kg, 91cm and12.3 respectively; all below the third percentile. She had good volume synchronous pulses with a rate of 100/min and heart sounds that were soft and muffled. Her abdomen was distended with a girth of 56cm and an umbilical defect of 1cm. She was alert and easily distracted. There was no neck swelling. Her tone and deep tendon reflexes were normal

An assessment of congenital hypothyroidism with pericardial effusion was made. The thyroid function test showed markedly elevated TSH of > 50(µi.u/ml) and unrecordable levels of T4 and T3. The ultrasound of the neck revealed reduced thyroid size, right lobe 10.3 x 7.8mm and left 9.0 x 10.1mm (Figure 1), while echocardiography showed mild pericardial effusion. The x-ray of the wrists/knees showed gross demineralization with only 2 carpal bones ossified. The patella was not ossified and there were growth arrest lines (Figure 2). Her bone age was that of a 2yr old.

Visual and hearing assessments were essentially normal. She also had mild normocytic, normochromic anaemia (PCV 25%). She was commenced on L-thyroxine therapy initially at 25µg daily and gradually increased to 4µg/kg daily. Over a follow up period of six years (till 11 years), her growth velocity increased moderately with weight and height for age between the 5th and 10th percentile, T3 and T4 normalized (Table 1), skin became smooth and appetite improved. Her vision, speech, reading and writing improved dramatically. She was doing better in school, although she was 2 classes behind her peers.

Table 1: Serial Thyroid assays and anthropometry.

Parameter	Presentation	9months	18months	36months	54months	72months
TSH(ui.u/ml)	>50	0.1	3.3	1.1	0.5	16.8
T3 ng/ml	0.0	1.0	1.4	1.4	1.3	1.1
T4 ug/dl	0.0	11.6	10.3	14.2	5.0	9.9
WEIGHT(kg)	10	15	17	21	21.5	27
HEIGHT(cm)	91	100	105	118	124	134
BMI	12.1	15	15.4	15.1	13.9	15.1

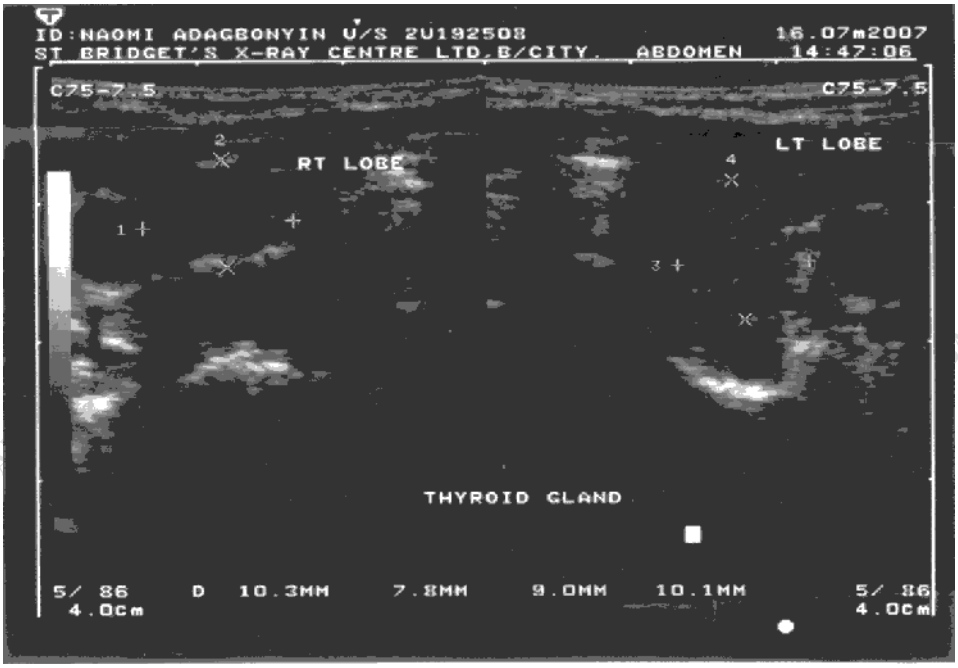

Figure 1: Ultrasound of the thyroid gland.

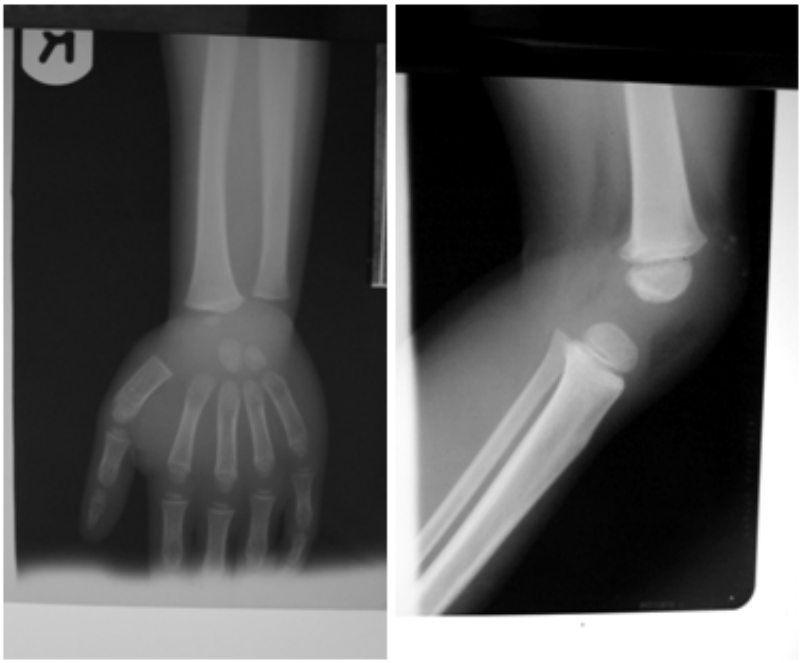

Figure 2: Radiograph of the knee and hand

DISCUSSION

The estimated prevalence of congenital hypothyroidism (CH) in Nigeria is 42,500. The aetiologies include: Thyroid dysgenesis (75%), Thyroid dyshormonogenesis (10%), Transient hypothyroidism (10%) and Hypothalamic-pituitary (TSH) deficiency (5%) (Shah, 2004). The congenital hypothyroid child may appear normal at birth or present with slightly increased head size due to myxedema of the brain. There may be prolongation of physiological jaundice, lethargy, somnolence, large tongue and nasal obstruction. Affected infants cry little, sleep more and are very lethargic. There may be presence of umbilical hernia, hypothermia, constipation, edema of genitals and extremities, cardiomegaly, bradycardia and asymptomatic pericardial effusion (Gomella *et al*., 2013; Le-Franchi, 2011; Shah, 2004).

Growth stunting and poor mental development manifest during infancy. However, the onset and severity of symptoms depend on the aetiology. Ideally a neonatal screening for CH should be routinely done in all children as treatment of affected infants within six weeks of birth leads to normal mental and physical development (Le-Franchi, 2011; Kayode-Adedeji, 2015; Hindmarsh, 2002; Hopwood, 2002). Delay in diagnosis and treatment can cause severe, irreversible mental and physical retardation. (Mahmoud, 2013; Le-Franchi, 2010). The diagnosis is made by an assay of the thyroid hormones, showing elevated TSH and low T4 and T3 (Le-Franchi, 2011; IAEA, 2005; Toblanc, 1999).

Most children with CH who are correctly treated with thyroxine grow and develop normally in all respects. Even most of those with athyrosis and undetectable T4 levels at birth develop with normal intelligence. Few treatments in the practice of medicine provide as large a benefit for as small an effort, when the cost of treatment is compared to the profound clinical impact. (Shah, 2004). The goal of therapy is early adequate thyroid hormone replacement. It is desirable to maintain the serum T4 in the upper half of normal range in infants; an initial dose of 10-15µg/kg/day in neonates and young infants, while older children may require 4µg/kg/day (Shah, 2004; Hindmarsh, 2002; Hopwood, 2002).

In developed countries, where neonatal screening is done routinely, the prognosis for affected infants has improved dramatically. However this is not available in Nigeria, so her diagnosis was not made at birth. Furthermore, in the case highlighted, she appeared normal to her parents until about two years of age when she practically stopped growing and subsequently could not read or write like her peers. This patient unlike most others did not have obvious features in infancy; the delay in overt manifestation in this case is due to the transplacental transfer of moderate amounts of maternal T4, coupled with the function of the hypoplastic gland. This late onset of symptoms is in keeping with thyroid gland hypoplasia. The outlook for patients whose onset is after two years is much better even in the face of delay in diagnosis and treatment. In this patient, though the growth velocity, vision and school performance improved, she is unlikely to fulfill her potentials physically and mentally.

The consequences of untreated CH, particularly severe mental retardation and growth restriction, constitute a great burden to the family and nation at large. The cost of special education and institutionalization of affected children is at least 10-fold the cost of the screening programme.

CONCLUSION

The index of suspicion of congenital hypothyroidism in developing countries is low when evaluating children with failure to thrive; this case emphasizes the need for a screening programme for hypothyroidism in developing countries. Early diagnosis and prompt treatment can make the difference between a genius and an institutionalized severely retarded individual

ACKNOWLEDGEMENT

The authors acknowledge the contribution of the radiologist -Dr Eluehike. S. in the evaluation of the radiologic investigations of the patient.

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AUTHORS CONTRIBUTIONS

Kayode-Adedeji, B.O was involved in manuscript writing and literature review, while Alikah, S.O. was involved in manuscript writing.