CONGENITAL HYPERTHYROIDISM WITH DOWN SYNDROME WITH MICROCEPHALY AND GROSS DEVELOPMENTAL DELAY
(A Rare Case Report)
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Abstract
Attempts have been made to report the rare occurrence of congenital hyperthyroidism with Down Syndrome as the same is commonly associated with congenital hypothyroidism. The baby admitted to our hospital for diarrhea but the parents were ignorant about the syndromic nature of the baby. After doing thorough work out of the case the child found to be suffering from Down syndrome. As Down syndrome is commonly associated with congenital hypothyroidism & the clinical presentation also supports the same ,we sent blood sample for thyroid profile but surprisingly the results put all of us in dilemma as report found to be hyperthyroidism .We sent blood sample again but the results were same.

Key words: Down Syndrome(DS), Hypothyroidism, Hyperthyroidism, Karyotyping, Head circumference(HC), Microcephaly, Developmental Delay.

INTRODUCTION

Congenital hyperthyroidism is a very rare disease. But, for each affected child it has to be considered as a serious condition because of the negative impact of hyperthyroidism on fetal and postnatal development[1]. Neonatal hyperthyroidism occurs in only about 2% of infants born to mothers with a history of Graves disease[2,3]. It affects boys as often as girls[3]. Hyperthyroidism is less frequent in patients with Down syndrome i.e.,between 0.8% and 2.5%[4]. Prematurity is frequent, as well as hypotrophia. Tachycardia, cardiac arrhythmia, goiter, hyperexcitability, poor weight gain, growth retardation, hepatomegaly and/or splenomegaly, stare and/or eyelid retraction are among the most frequent neonatal thyrotoxicosis clinical signs[5]. The main complication of persistent hyperthyroidism in the neonatal period and during infancy is craniosynostosis[2]. Most cases are due to maternal antibodies transferred from the mother into the fetal compartment, which stimulate the fetal thyroid by binding to the thyrotropin (TSH) receptor. To confirm the immune nature of hyperthyroidism, thyroid-stimulating immunoglobulins (TSI) should be determined[5].

PATIENT AND METHOD:
Term 6m MCh (Fig-1) presented with fever, poor weight gain, diarrhoea with family h/o Graves disease in maternal grand father and no significant drug history or thyroid or other endocrinal diseases in mother. The child has not attend head control & smiling at mother. Anthropometry revealing weight of 5800g, length of 63.8cm, HC of 38.8 cm. Anterior fontanel normal in shape and size. Posterior fontanel closed. General examination revealing features of of DS (low set ear, upward slanting palpebral fissure, single palmar creuse, kennedy’s line, protruded tongue, high arch palate)(Fig-1/2), coarse skin, pallor, icterus, tachycardia (124-140 bpm) & fever(100.4°F). Systemic examination revealing hepatosplenomegaly.

On routine haemogram Hb is 8.8gm%(Sahali’s hemoglobinometer), Thyroid profile (fT₃=660 pg/dl, fT₄=6.2ng/dl, T3=33.2ng/dl, T3=280ng/ml, TSH=1.01µU/ml, CLIA method); Karyotyping revealing Trisomy 21(GTG Banding)(Fig-3).
RESULT AND DISCUSSION:
The baby is found to be having microcephaly (HC <3rd percentile, WHO)[6], underweight (wt for age is < 3rd percentile, WHO)[7] and gross developmental delay (both in gross motor and social aspect). Thyroid profile revealing hyperthyroidism[8] and Trisomy 21 revealing DS[9]. Gross developmental delay are usually associated with congenital hypothyroidism[7] but it is unusual with hyperthyroidism as in this case. Congenital hypothyroidism is having normal or increased head size(macrocephaly)[7] but here there is microcephaly. Anemia is more common in congenital hypothyroidism[10] than congenital hyperthyroidism.

CONCLUSION:
Most cases resolve spontaneously by 3rd to 4th month of life [5]. Early diagnosis & t/t favours good outcome[11]. Advanced osseous maturation, microcephaly & mental retardation may occur if t/t is delayed. Mortality rate varies from 16% to 25% mostly due to cardiac failure &/or dyselectrolytemia[1]. Congenital hyperthyroidism is more prevalent in patients with DS than in the general population. It has no gender predominance. It is caused mainly by Graves’ disease. Anti-thyroid drugs were not effective in achieving remission and radioactive iodine as a definitive treatment was required in all cases. The case presented because of its rare presentation & diagnosis at our hospital but unfortunately lost follow up.

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