CASE REPORT

Congenital Hypothyroidism Associated with Maternal Hypothyroidism and Iodine Deficiency During Pregnancy

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Abstract:
Congenital hypothyroidism is a partial or complete loss of function of the thyroid gland (hypothyroidism) that affects infants from birth (congenital). The thyroid gland is a butterfly-shaped tissue in the lower neck. It makes iodine-containing hormones T3 and T4 that play an important role in regulating growth, brain development, and the rate of chemical reactions in the body (metabolism). People with congenital hypothyroidism have lower-than-normal levels of these important hormones. We present a case of a baby born with congenital hypothyroidism whose mother developed iodine deficiency during pregnancy due to use of non-iodized salt and lack of proper prenatal care.

Key words:
Congenital hypothyroidism, iodine deficiency, maternal iodine deficiency

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Received date: 10/07/2017 Revised date: 08/12/2017 Accepted date: 10/12/2017

DOI Link: http://doi-ds.org/doilink/12.2017-13697469/

Case presentation:
A baby boy was found to have macroglossia, severe constipation and lethargy within few days after birth (39 weeks gestational age with normal vaginal delivery) in rural India in Konkan region (Ratnagiri, Maharashtra). About 1 month after birth the TSH levels were checked and were very high TSH-150 (0.5-4.5 mIU/L), with low T4 and T3. The baby was started on levothyroxine 25 mcg daily and it resolved the constipation, lethargy and poor growth. On further review it was evident that the mother (25 years primi-gravida) was not given pre-natal vitamins during the pregnancy. Instead of using iodized salt in cooking she was asked to use saindhav (rock salt) by family due to improper advice by a local natural physician. Baby had a thyroid bed US done which showed absence of thyroid gland. Family was advised to do a radioactive iodine uptake and scan but due to lack of facility in the local area and fear about the word “Radioactive” family did not do it. On work up mother was also found to have hypothyroidism and was also started on levothyroxine after delivery. Now baby is 4.5 years old and is stable on levothyroxine 37.5 mcg daily and is showing normal physical and mental development.

Discussion:
Congenital hypothyroidism (CH), one of the most prevalent endocrine diseases, is known as a common preventable cause of mental retardation. The prevalence of CH in India is 1 in 2640 based on the study was done by Desai et al. in 1998. The other studies from India quote a prevalence of 1 in 1985 from Hyderabad and 2.1 in 1000 from Kochi. A study by Kishore KR, et al revealed that CHT in India has a high incidence and is an URGENT high priority for public screening. The first multi-centric study screening above 1 lakh neonates born throughout India was launched by Indian Council of Medical Research (ICMR) National Task Force Team on New Born Screening (NBS) at AIIMS New Delhi (2007–2012) and the preliminary results reveal a much higher incidence of CH all over India at 1 in 1172, particularly in south Indian population (1 in 727). Mass population screening of newborn infants for CH, first introduced in 1974, is today a routine and effective tool of timely/early diagnosis of CH, used in most of the world. Initiation of treatment within the first two to three weeks of life resulted in both normal IQ and physical growth. Many risk factors contribute to the etiology of CH. In particular, a multifactorial origin of CH in which genetic
(high frequency of additional malformations) and environmental factors (especially iodine deficiency and maternal diabetes) play a role in the development of the disease.\textsuperscript{(11,12)} The signs and symptoms of hypothyroidism include lethargy, coarse facial features, poor feeding or weight gain, jaundice, hoarse cry, macroglossia, large fontanelles, umbilical hernia, cool, dry skin, myxedema, goiter, constipation.\textsuperscript{(13)} But the physical finding may not be present at birth. Elevated TSH levels with low free T4 levels are suggestive of hypothyroidism. In humans, untreated congenital hypothyroidism due to thyroid agenesis inevitably leads to cretinism, which comprises irreversible brain dysfunction and dwarfism. The appropriateness of the recommended L-thyroxine dose (10-15 $\mu$g/kg/day) for the treatment of congenital hypothyroidism has been questioned because of the risk of iatrogenic hyperthyroidism. Average range IQ scores and positive behavioral adaptation are observed in congenitally hypothyroid children treated with L-thyroxine doses lower than currently recommended; the L-thyroxine dose should be individualized to prevent iatrogenic hyperthyroidism. TSH normalization should not be a primary objective of treatment.\textsuperscript{(14)} Iodine deficiency is a major public health problem; globally, it is estimated that two billion individuals have an insufficient iodine intake. Although goiter is the most visible sequelae of iodine deficiency, the major impact of hypothyroidism due to iodine deficiency is impaired neurodevelopment, particularly early in life. In the fetal brain, inadequate thyroid hormone impairs myelination, cell migration, differentiation and maturation. Moderate-to-severe iodine deficiency during pregnancy increases rates of spontaneous abortion, reduces birth weight, and increases infant mortality. Offspring of deficient mothers are at high risk for cognitive disability, with cretinism being the most severe manifestation. It remains unclear if development of the offspring is affected by mild maternal iodine deficiency. Moderate-to-severe iodine deficiency during childhood reduces somatic growth.\textsuperscript{(15, 16)} In-spite of that there are some people who are using non-iodized salt in cooking and keeping the pregnant women away from modern health care and pre-natal care due to ignorance and misguidance.

Conclusion:

This case stressed the importance of having high index of suspicion for congenital hypothyroidism in babies with lethargy and failure to thrive. It is important to diagnose these babies early in life and treat to prevent mental retardation and cretinism. Maternal
pre-natal care using multivitamin containing 225 mcg of iodine and use of iodized salt in cooking should be stressed.

Conflict of interest: None to declare
Source of funding: Nil

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