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A Missing Thyroid! In an Infant: A Case Report

Dr. Ponnas Janakiraman[1], Dr Ajay Raj[2], Dr KK Choudhary[3], Dr Kalpak Mondal[4], Dr Ravi Varma[5]

[1]JR III, Paediatric Medicine, Mahatma Gandhi Memorial Medical College & Hospital, Jamshedpur
Corresponding Author Email: *ponnas93[at]live.in*

[2]Professor & HOD, Paediatric Medicine, Mahatma Gandhi Memorial Medical College & Hospital, Jamshedpur

[3]Professor, Paediatric Medicine, Mahatma Gandhi Memorial Medical College & Hospital, Jamshedpur

[4]SR II, Paediatric Medicine, Mahatma Gandhi Memorial Medical College & Hospital, Jamshedpur

[5]SR II, Paediatric Medicine, Mahatma Gandhi Memorial Medical College & Hospital, Jamshedpur

Abstract: ***Thyroid dysgenesis: Aplasia, hypoplasia, or an ectopic gland is the most common cause of permanent congenital hypothyroidism, accounting for 80 - 85% of cases. In 66% of infants, rudiments of thyroid tissue are found in an ectopic location, anywhere from the base of the tongue (lingual thyroid) to the normal position in the neck (hypoplasia). In 33% of cases of dysgenesis, shows aplasia. Thyroid dysgenesis has a 2: 1 female: male ratio. This case reports a six months old female infant born to a non consanguineous parents shows a delay in developmental milestones and not gaining adequate weight with chronic constipation. Child shows a vital parameter of relative bradycardia and mild pallor, mild wasting and severe stunting with microcephaly and peculiar features of cretinoid facies with umblical hernia and dry and coarse skin. Peripheral blood smear reveals Macrocytic normochromic picture with normal WBC and platelet count, with Free T4 level of 0.08 ng/dl and TSH level of >50.2 mIU/L. USG report of neck reveals an absent thyroid gland. Hence this child got administered with levothyroxine (L - T4) at a dose of 67.5mcg OD in morning before breastfeeding and the child was periodically followed up in regular OPD basis.***

Categories: Endocrinology/Thyroid gland/Paediatric Medicine

Keywords: Thyroid dysgenesis, Congenital hypothyroidism

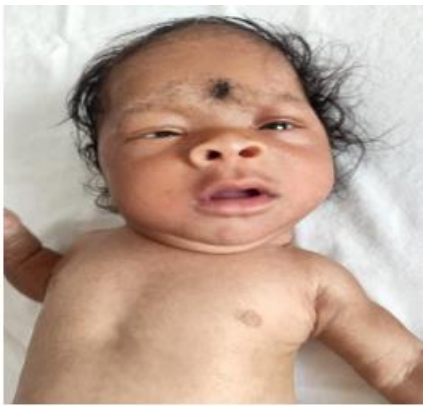
1. Introduction

Thyroid gland arises from an outpouching of the foregut at the base of the tongue (foramen caecum). The bilobed shape thyroid is recognized by 7th week of gestation, it migrates to its normal location over the thyroid cartilage by 8th – 10th week of gestation and characteristics thyroid follicle cell and colloid formation is seen by 10th week. Thyroxine (T4) and Triiodothyronine (T3) synthesis and secretion occurs by 12th week of gestation (1). Iodination of tyrosine forms monoiodotyrosine and diiodotyrosine; 2 molecules of diiodotyrosine couple to form 1 molecule of T4, or 1 molecule of diiodotyrosine and 1 molecule of monoiodotyrosine to form T3. Once formed, hormones are stored as thyroglobulin in the lumen of the follicle (colloid) until ready to be delivered to the body cells. Approximately 70% of the circulating T4 is firmly bound to Thyroid - binding globulin (TBG). Less - important carriers are transthyretin, and albumin. Only 0.03% of T4 comprises free T4. Approximately 50% of circulating T3 is bound to TBG, and 50% is bound to albumin; 0.30% of T3 comprises free, T3. (2) The thyroid is regulated by TSH, a glycoprotein produced and secreted by the anterior pituitary. TSH synthesis and release are stimulated by TRH, which is synthesized in the hypothalamus and secreted into the pituitary. (3)

2. Case Report

A six months old female infant came with chief complaints of Constipation / Not gaining weight/ Delay in developmental milestones, She was born through normal vaginal delivery in home with no significant history of perinatal asphyxia or NICU admissions, She was exclusively breastfeed up to 6 months and she was immunized up to date, She shows a significant delay in developmental milestones and she was born to non consanguineous parents in the birth order of 3 whose medical history was insignificant. Child shows a vital parameters relative bradycardia and mild pallor, mild wasting and severe stunting with microcephaly with wide & open anterior fontanelle measuring 4×4cms and shows a characteristic cretinoid facies (Sparse eyebrows, Narrow palpebral fissure, Depressed nasal bridge, Wide open mouth, Macroglossia and Tongue tie), low set ears, short and wide neck, umbilical hernia, dry and coarse skin. Other systemic examinations are within normal limit. Following Investigations like CBC shows Macrocytic normochromic anemia with normal WBC and platelet count and Thyroid profile shows Free T4 level of 0.08 ng/dl and TSH level of >50.2mIU/L.

Ultrasound report of neck reveals Thyroid agenesis.



3. Discussion

The cause of thyroid dysgenesis is unknown in most cases. Thyroid dysgenesis occurs sporadically, but familial cases occasionally have been reported. The finding that thyroid developmental anomalies, such as thyroglossal duct cysts and hemiagenesis, are present in 8 - 10% of 1st - degree relatives of infants with thyroid dysgenesis supports an underlying genetic component. Hypothyroidism results from deficient production of thyroid hormone, either from a defect in the gland itself (primary hypothyroidism) like dysgenesis, dyshormonogenesis, Maternal ab: Thyrotropin receptor–blocking antibody (TRBAb) Iodides, Medication like: Iodides, Amiodarone, Propylthiouracil, Methimazole & Radioiodine. Iodine deficiency (endemic goiter). Or due to reduced thyroid - stimulating hormone (TSH) stimulation (central or hypopituitary hypothyroidism) like Mutation of TSH β - subunit gene, TRH gene, PIT 1 mutation and PROP - 1 mutation. This may be manifested from birth (congenital) or acquired. At birth, there is an acute release of TSH; peak serum concentrations reach 60 mIU/L 30 min following delivery in full - term infants. A rapid decline occurs in the ensuing 24 hr and a more gradual decline over the next 5 days to <10 mIU/L. Serum TSH level in infancy are 0.5 - 5.5 mIU/L Serum free T4 levels are 0.9 - 2.3 ng/dL in infancy and decline to 0.7 - 1.8 ng/dL in childhood. Serum free T3 levels are 180 - 760 pg/dL in infancy and decline to 230 - 650 pg/dL in childhood. (4) Thyroid ultrasound examinations can determine the location, size, and shape of the thyroid gland, and they are useful for assessing the solid or cystic nature of nodules. Thyroid scanning may be indicated to assess the presence of thyroid tissue in questions of thyroid dysgenesis and to detect ectopic thyroid tissue, and thyroid uptake may be indicated to evaluate possible “hot” thyroid nodules. Diagnostic studies should be performed with 99mTc pertechnetate or 123I because they have the advantages of lower radiation exposure and high - quality scintigrams. (5)

4. Conclusion

Management of these conditions depends upon administration of levothyroxine (L T4) and periodical follow up. The recommended initial starting dose is 10 - 15 µg/kg/day and if patient has very low T4 (<5 µg/dL) use higher 12–17 µg/kg/24 hr dose.3–6 mon: 8–10 µg/kg/dose OD, 6 - 12 mon: 6–8 µg/kg/dose OD, 1–5 yr: 5–6 µg/kg/dose OD, 6–12 yr: 4–5 µg/kg/dose OD, >12 yr: Incomplete growth and pre - puberty: 2–3 µg/kg/dose OD, complete growth and puberty: 1.7 µg/kg/dose OD. Levels of serum T4 or free T4 and TSH should be monitored at recommended intervals: Every 1 - 2 month in the 1st 6 month of life, and then every 2 - 4 month between 6 month and 3 year of age. The goals of treatment are to maintain the serum Free T4 or total T4 in the upper half of the reference range for age, with serum TSH in the reference range for age, optimally 0.5 - 2.0 mIU/L. The dose of l - T4 on a weight basis gradually decreases with age. Care should be taken to avoid prolonged under - treatment or over - treatment.

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