Brief Communication

Cretinism revisited

Ankit Srivastav, Indira Maisnam, Deep Dutta, Sujoy Ghosh, Satinath Mukhopadhyay, Subhankar Chowdhury

Department of Endocrinology, Institute of Post Graduate Medical Education & Research, Kolkata, India

ABSTRACT

Objective: Cretinism is a condition of severely stunted physical and mental growth due to untreated congenital hypothyroidism. It has been largely eliminated in the developed world, though we still continue to see cases in India. **Case Report:** A 22-year-old male was brought to our Endocrine clinic by his brother due to his "not growing up." The patient was 83 cm in height (SDS – 16.98) and weighed 13.9 kg (<3rd percentile). He had dull look, puffy face with thick lips, macroglossia, and umbilical hernia. There was sexual infancy with prepubertal testes (<3 ml). He could sit without support, but could not stand, or walk without support and could only talk in monosyllables. He was born full term by normal vaginal delivery, and cried immediately after birth. The developmental milestones were delayed, and not achieved till date. He is the eldest of seven siblings, rest six of whom have no complaints. An X-ray of hand was done showing bone age of less than 1 year. A thyroid profile showed TSH >150 IU/ml, free T4 and T3 below the assay range. Ultrasound of neck showed absent thyroid tissue in neck. Iodine-131 uptake scan was consistent with thyroid aplasia. Diagnosis was myxematous cretinism due to thyroid aplasia was made, and patient was started on thyroxine supplementation. **Conclusion:** This case represents the most severe form of untreated congenital hypothyroidism presenting as severely stunted physical and mental growth with delayed bone and sexual maturation.

Key Words: Cretinism, thyroid aplasia, iodine 131 uptake

Introduction

Cretinism is a condition of severely stunted physical and mental growth due to untreated congenital deficiency of thyroid hormones (congenital hypothyroidism). Congenital hypothyroidism can be endemic, genetic, or sporadic. Endemic cretinism was especially common in areas of southern Europe around the Alps, Bangladesh, China, and Nepal. Sporadic and genetic cretinism results from abnormal development, or function of the foetal thyroid gland.[1] However, cretinism has been almost completely eliminated in developed countries by early diagnosis by newborn screening schemes and iodine supplementation programs.

Access this article online	
Quick Response Code:	Website: www.ijem.in
	DOI: 10.4103/2230-8210.104081

Case Report

A 22-year-old male was brought to our Endocrine clinic by his brother due to his "not growing up." The patient was 83 cm in height (SDS – 16.98) and weighed 13.9 kg (<3rd percentile). He had dull look, puffy face with thick lips, macroglosia, and umbilical hernia. There was sexual infancy with prepubertal testes (<3 ml). He could sit without support, but could not stand or walk without support, and could only talk in monosyllables. He was born full term by normal vaginal delivery, and cried immediately after birth. The developmental milestones were delayed and not achieved till the date. He is the eldest of seven siblings, rest six of whom have no complaints. An X-ray of hand was done showing bone age of less than 1 year. A thyroid profile showed thyroid stimulating hormone (TSH) >150 IU/ml, free T4 and T3 were below the assay range. Ultrasound of neck showed absent thyroid tissue in neck. Iodine-131 uptake scan was consistent with thyroid aplasia. Diagnosis was myxematous cretinism due to thyroid aplasia was made, and patient was started on thyroxine supplementation.

Corresponding Author: Dr. Shrivastav Ankit, Department of Endocrinology, SSKM Hospital, 244, AJC Bose Road, Kolkata - 700 020, India. E-mail: ankit.med@gmail.com

Discussion

This case represents the most severe form of untreated congenital hypothyroidism presenting as myxedematatos cretinism along with mental retardation.[2]

Cretinism may be of two types: Neurological or myxedematatos.

The three characteristic features of neurological endemic cretinism in its fully developed form are extremely severe mental deficiency together with squint, deaf mutism, and motor spasticity. They usually have a goiter. The neuropathological basis of the clinical picture includes under-development of the cochlea for deafness, maldevelopment of the cerebral neocortex for mental retardation, and maldevelopment of the corpus striatum (especially putamen and globus pallidus) for the motor disorder. The cerebellum, hypothalamus, visual system, and hippocampus are relatively spared. Neurological cretinism is now thought to be predominantly caused by maternal hypothyroidism due to iodine deficiency. It may have an autosomal recessive predisposition also.

Myxedematatous cretinism may present with severe growth retardation, incomplete maturation of the facial features, including the naso-orbital configuration, atrophy of the mandibles, puffy features, myxedematous, thickened and dry skin, dry and decreased hair, eyelashes and eyebrows, and much delayed sexual maturation. Goiter is usually absent and the thyroid is often not palpable, suggesting thyroid atrophy. Thyroidal uptake of radioiodine is much lower than in the general population. The serum levels of T4 and T3 are extremely low, often undetectable, and TSH is dramatically high. Markedly enlarged sella turcicae have been demonstrated.[3]

The morbidity from congenital hypothyroidism can be reduced to a minimum by early diagnosis and treatment. Most developed countries have neonatal screening tests in place.

Thyroxine must be dosed as tablets only, even to newborns, as the liquid oral suspensions, and compounded forms cannot be depended on for reliable dosing. In the case of dosing infants, the T4 tablets are generally crushed and mixed with breast milk, formula milk, or water.[4] Frequent monitoring (every 2-3 weeks during the first months of life) is recommended to ensure that infants with congenital hypothyroidism remain within the high end of normal range.

References

1. Chen ZP, Hetzel BS. Cretinism revisited. Best Pract Res Clin Endocrinol Metab 2010;24:39-50.
2. LaFranchi S. Congenital hypothyroidism: Etiologies, diagnosis, and management. Thyroid 1999;9:735-40.
3. Jalil MQ, Mia MJ, Ali SM. Epidemiological study of endemic cretinism in a hyperendemic area. Bangladesh Med Res Counc Bull 1997;23:34-7.
4. Eastman CJ, Zimmermann M. The iodine deficiency disorders. Thyroid Disease Manager. Ch. 20., 2009.

Cite this article as: Srivastav A, Maisnam I, Dutta D, Ghosh S, Mukhopadhyay S, Chowdhury S. Cretinism revisited. Indian J Endocr Metab 2012;16:S336-7.

Source of Support: Nil, **Conflict of Interest:** None declared