

## Congenital Hypothyroidism Diagnosed At 9 Years Old

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### Abstract

Congenital hypothyroidism is a common endocrine disorder of newborn. It is an important cause of global developmental delay. The early diagnosis and treatment is crucial. In this paper we are describing case of congenital hypothyroidism in Saudi Arabia presenting as 9-year-old Bormann boy who was born at home with no neonatal screening and family ignorance. He was discovered incidentally with chest infection, despite of that screening for congenital hypothyroidism has been available in since 1989. This case shows the importance of conducting newborn screening for early detection of congenital hypothyroidism in newborn, which is still being neglected in some developing countries. It also shows the importance of early starting adequate replacement therapy to prevent irreversible sequels.

### Key words

Congenital; Hypothyroidism; Developmental Delay

### Introduction

Congenital hypothyroidism is one of the important preventable causes of global developmental delay. Prevalence of newborn cases diagnosed worldwide is 1:3500 to 1:5000 [1]. In Saudi Arabia, the incidence is 1:2500, which is considered one of the highest where majority of cases are sporadic [1]. Newborn screening for congenital hypothyroidism has become routine since 1989 in Saudi Arabia. It instead of <sup>th</sup>rd<sup>th</sup> the population especially the mothers about the clinical symptoms of congenital hypothyroidism such as decreased activity, constipation and poor feeding. Advise them that the earlier the detection, the better the prognosis. There are main studies of value about the Congenital hypothyroidism in Saudi Arabia, according to study carried out from 1985 to 2000 in the capital city Riyadh, screened one hundred and twenty-one thousand, four hundred and four infants over a period of 15 years. The research conclude that the initial measurement of TSH in cord blood captures 97% of infants and the incidence of Congenital hypothyroidism was 1:2759 live birth with female: male ratio of 2:1 [2]. Also a second large study published in 2011 including the screening of 1007350 newborns in king Khaled hospital, reveled congenital hypothyroidism in 306 indicating an incidence 1 in 3292. So the incidence of congenital hypothyroidism in our country is higher than that reported from Europe and America,

with regional variation in incidence due to high consanguinity rate and multiple siblings involved in the families [1]. Unfortunately, due to lack of national health register including the statistics of the number of how many infants are screened, and how many not screened nationally each year. We really can't till the population statistics regarding congenital hypothyroidism.

### Case Presentation

In this case report we are describing 9-year-old boy with congenital hypothyroidism, which was neglected, not diagnosed and left untreated due to unawareness of his parents.

Nine years old Bormann boy, lives in Saudi Arabia, was brought to the Emergency Department for the first time with bronchopneumonia.

He was born at home to a family with low socio-economic status. He was not screened for hypothyroidism and never received any vaccination. This is the first time the mother approach for medical advice. No family history of thyroid disease or dysmorphogenesis with positive consanguinity

Systemic examination were normal apart from signs of chest infection, he was noticed to have coarse facial features, myxedematous face, prominent eyebrows, macroglosia, dry brittle hair, dry thick skin and diffuse goiter (Figure 1). He is on nasogastric tube due to difficulty in swallowing of solid food. Anthropometric measurements were as following: head circumference of 48cm with wide open anterior fontanel 2x3 cm, height 85 cm (SDS-8.69), upper segment 55 cm, and lower segment 30 cm, arm span 64 cm, which shows a disproportionate short stature. Weight was 8 kg (SDS-5.35). Higher brain function assessment revealed severe mental deficiency



Figure1: 9-year-old boy with congenital hypothyroidism. Showing coarse facial features; myxedematous face, prominent eyebrows, macroglosia, dry brittle hair, dry thick skin and diffuse goiter and frog like position.

He has generalized hypotonia with frog like position and hyporeflexia. Developmentally he has global delay, he rolls from side to side, sits with support, holds objects but can't transfer from hand to hand, only cooing, with no social interaction, and thus his developmental age was 6 months at presentation. He was investigated for congenital hypothyroidism was his TSH was 100 mIU/L (0.27-4.2), pmol/L (12-22). The radionuclide imaging showed thyroid agenesis

but clinical re-examination revealed he has a goiter which could be explained by autosomal recessive dyshormonogenesis combined with a poor quality scan. The diagnosis of severe iodine deficiency was considered rarely the absence of the risk factors. Then he was started on thyroxin replacement therapy, dose of (25mcg). Dose was gradually increased dependably on follow-up thyroid function tests, currently he is on 50 mcg once daily.

## Discussion

Late diagnosed congenital hypothyroidism has different clinical presentations, and it's uncommon to be presented at late childhood due to wide spread of neonatal screening. Our index case was presented with global developmental delay, severe mental deficiency and thyroid swelling. In literature there were reports with late presentation beyond 5 years old, the most common presentations were delayed growth, mental sub-normality, lethargy, chronic constipation or thyroid swelling [3,4]. Another different presentation was reported in 17 years old female with a delayed puberty and short stature [5]. Although menorrhagia is a frequent presentation of hypothyroidism, it's rare with congenital hypothyroidism. It has been reported in adulthood life and some of those patients were presented only with anemia [4,6].

## Conclusion

We are describing a case of late diagnosed congenital hypothyroidism in 9 –year-old boy.

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