Delayed Diagnosis of Congenital Hypothyroidism in a 6-month-old Male Infant in Tamale, Ghana.

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INTRODUCTION 1

- Congenital hypothyroidism is a common inborn endocrine disease.
- There have been reported increase in the prevalence of congenital hypothyroidism because of improvement in neonatal medicine and inborn screening programs, as well as increase in sensitivity of testing methods.
- In Africa, studies in Nigeria have shown varying incidence between 16.4% and 1.4%. However, a lower incidence (0.1%) was reported in South Africa.
- In Ghana, not much has been done to ascertain the incidence of this disorder, and the country lacks newborn screening program for this disorder. However, there has been a case report in a tertiary facility in Ghana which was also missed in the neonatal period.

(Grasberger & Refetoff, 2011), (Dayal & Prasad, 2015), (Rastogi & LaFranchi, 2010), (Bona et al, 2015), (Deladoëy & Van Vliet, 2014), (Otokunefor et al,2017), (Ameyaw & Ameyaw, 2017)

INTRODUCTION 2

- Majority of newborns with congenital hypothyroidism may be asymptomatic at birth, making newborn screening the most certain way for early diagnosis screening .
- In symptomatic cases, presentation may include poor feeding, poor growth, hoarse cry prolonged sleep and constipation, jaundice and macroglossia.
- On physical examination, there may be widening of the fontanelles, large protruding tongue, dry skin, umbilical hernia and delayed reflexes.
- We present a case of delayed diagnosis, although he had a number of warning signs in the neonatal period

(Dayal & Prasad, 2015), (Rastogi & LaFranchi, 2010), (Ameyaw & Ameyaw, 2017)

CASE PRESENTATION 1

- Our case is a 3 year, 4 months old male, who was first seen at our pediatric outpatient clinic at 5 months of age on account of poor growth. He was delivered via elective C/S on account of two previous C/S deliveries.
- The birth weight was 3.1 kg. He **passed meconium after day four of life** and subsequently was able to pass stool at least once every day.
- According to the mother, **he developed jaundice in the first week** of life which was managed at home by exposure to early morning sun for two days. The mother also recalled that the baby used to sleep a lot during the neonatal period.

CASE PRESENTATION 2

- At the initial presentation, he had **coarse faces and a protruding tongue, widened anterior fontanelle and herniation of the umbilicus**.
- The neurological assessment at presentation revealed good neck control which was achieved after **three months of age, inability to sit with support, general hypotonia in both ventral and vertical suspension maneuvers and weak palmar grasp reflex.**
- The examination findings of the other systems were essentially normal.

CASE PRESENTATION 3

- Provisional diagnosis of congenital hypothyroidism was made based on the initial assessment.
- Diagnosis was confirmed with abnormal thyroid function test : **TSH 172.832 uIU (0.38-5.33); FT4: <5.1 pmol/L (7.9-18.5); FT3: 1.7 pmol/L (3.5-7.8)**
- Anterior neck ultrasonography revealed orthotopic thyroid gland of normal size and no abnormalities detected.

CASE PRESENTATION 4

- He was started on oral levothyroxine 50 microgram daily and scheduled for clinical review with repeat thyroid function test results after six months.
- At the first follow-up visit, patient was able to sit with support but had truncal hypotonicity. The repeat thyroid function test results at this visit wase as follows: TSH, 2.186 uIU (0.38-5.33); FT4: 16.6 pmol/L (7.9-18.5); FT3: 6.8 pmol/L (3.5-7.8).
- As the laboratory parameters returned to normal levels, the treatment was continued at same dose of levothyroxine and a plan to review the patient every six months in order to monitor growth and development and to adjust levothyroxine levels when necessary.
- However, patient defaulted for review due to family issues, and there were irregularities in the administration of the levothyroxine.

CASE PRESENTATION 5

- After two years of interruption without follow-up, the child was seen with the mother at our clinic when he was now three years four months old.
- He was stunted and underweight after assessment. The mother reported that he started walking at 3 years of age and she admitted that patient has significant improved following the thyroid hormone replacement therapy.
- The patient also had delayed speech as he could only utter a few discernible words such as ‘mama’
- The patient’s medication was adjusted appropriately and the family was referred for nutritional counseling and rehabilitation and speech therapy

CONCLUSION

- In conclusion, this case highlights the importance of early diagnosis and initiation of therapy to avoid complications in both physical and mental development associated with congenital hypothyroidism.
- It also highlights the importance of regular follow-up to ensure optimal care after diagnosis and initiation of therapy.
- Universal newborn screening remains the standard for early diagnosis of this condition but in its absence, caregivers should be educated on the early neonatal period and perinatal and neonatal care should have a high level of suspicion.

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