Late Diagnosis of Congenital Hypothyroidism in Young Adult

Laurentius A. Pramono, Angelina Yuwono

Department of Internal Medicine, Faculty of Medicine, Atma Jaya Catholic University of Indonesia, Atma Jaya University Hospital, Jakarta, Indonesia.

Corresponding Author:
Laurentius A. Pramono, MD. Department of Internal Medicine, Faculty of Medicine, Atma Jaya Catholic University of Indonesia, Atma Jaya University Hospital. Jl. Pluit Selatan Raya no. 19, Penjaringan, Jakarta 14440, Indonesia. email: l_aswin@hotmail.com.

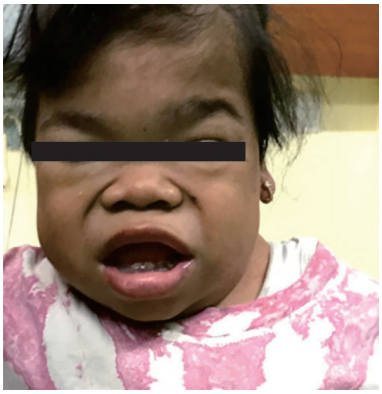

Figure 1. Myxedematous face and macroglossia

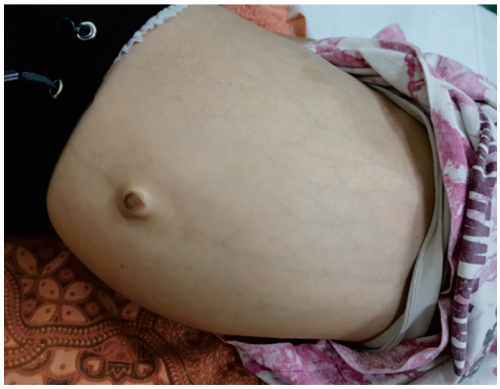

Figure 2. Umbilical hernia

Figure 3. Short stature

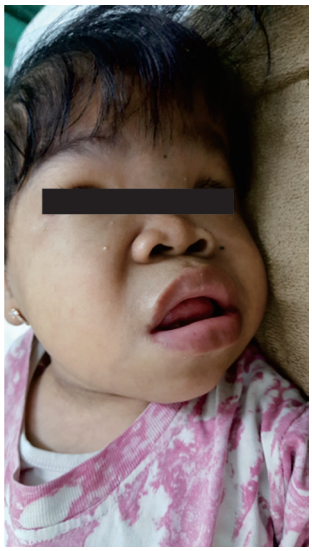

Figure 4. Hypotonic posture and macroglossia

Congenital hypothyroidism is the most treatable cause of mental retardation.[1] It is also the most prevalent congenital endocrine disorder in childhood. A dramatic improvement can be made by early detection, diagnosis, and adequate treatment with levothyroxine in patients with congenital hypothyroidism.[2] Severe cognitive impairment is associated with persistent disease in patients who have delayed or no treatment at all.[1,2] In this modern era with complete healthcare facilities in a big city like Jakarta, the prevalence of late-diagnosed congenital hypothyroidism is supposed to be very low. Since many districts have their own public healthcare facilities to screen and diagnose congenital hypothyroidism in children at very young age, a delayed diagnosis in adulthood is actually a rare case.

In this medical illustration, we report a case of 21 year-old woman who came to our hospital with abdominal pain. She had mental retardation with no capability to communicate well with other person. She had a short stature. She also had myxedematous face with big lips and a very big tongue. There was no goiter or lump on her neck. Her motoric performance was very weak and frail. During abdominal examination, we could see an umbilical bulging on her abdominal wall and on palpation, we could feel an umbilical hernia. By abdominal ultrasound, we could see the umbilical hernia. Unfortunately, no diagnosis of congenital hypothyroidism had been made when she was a newborn, there was also no past or known history of thyroid disease of her and her family. She had a diagnosis of mental retardation with no specific etiology since she was 5-years old.

Based on the results of her laboratory examination, we had a confirmed diagnosis of primary hypothyroidism with T4 10.56 nmol/L (normal 60-120 nmol/L) and TSH > 100 µIU/mL. We provided her treatment using levothyroxine based on her body weight (25 µg daily). We arranged her to have abdominal CT Scan and digestive surgery as further management for her umbilical herniation.

Some defects are correlated with congenital hypothyroidism when the disease is not treated properly and adequately.[3] Neurocognitive, neuromotoric, growth, and development are some areas which can be disrupted by long-term hypothyroidism condition for patients who had the disease since their early years of life.[3,4] Congenital hypothyroidism appears to be associated with an increased risk of congenital malformations. Several congenital malformations associated with congenital hypothyroidism are umbilical hernia, congenital heart disease, neurologic abnormalities, genitourinary malformations, cleft palate, and Down's syndrome.[3]

Studies concluded that severity of the congenital hypothyroidism has more important role than timing of treatment initiation on long-term cognitive and motor outcomes.[5] Detrimental effects on developmental outcomes in congenital hypothyroidism patients may persist over time; however, early treatment for patients at very early ages may bring the best cognitive outcomes and neuromotoric development.

Regardless of the treatment options, we can say that it is a loss case and a very late diagnosis and treatment of congenital hypothyroidism. The unusual age of detection, delayed diagnosis and treatment are some reminders for primary care physicians in our society to pay greater attention to screening programs.[6] Early detection and prompt treatment is an essential part of measures to reduce burden of mental retardation in our society. Delayed diagnosis of congenital hypothyroidism case, which is diagnosed at adulthood, indicates failure in screening program. Early diagnosis and treatment are necessary to prevent long-term catastrophic effects. This is a wake-up call for attention and awareness for society and primary care physicians in our country.[6]

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