



Primary Congenital Hypothyroidism in Children Below 3 Years Old - Etiology and Treatment With Overtreatment and Undertreatment Risks, a 5-Year Single Centre Experience

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Worldwide neonatal screening for congenital hypothyroidism (CH) is a gold standard of active surveillance in newborns. Prompt diagnosis, subsequent timely treatment implementation, and proper dosage of levothyroxine (L-T4) are crucial for normal growth and development, especially of the central nervous system. However, overtreatment may have a potential negative impact on further neurodevelopment. We retrospectively analysed data of 99 newborns with CH diagnosis, referred to the Endocrinology Outpatient Clinic of the Institute of Mother and Child in Warsaw, Poland from the CH screening program from 2017 to 2021. We evaluated the diagnostic process and treatment up to the age of 3 years. We compared groups of children from the first and the second screening groups (FSG, SSG) in the neonatal screening with an evaluation of ultrasound examination (thyroid dysgenesis vs. gland in situ, GtS). The overtreatment and undertreatment risks were assessed and an analysis of the new TSH thresholds was performed. Treatment was implemented at a median of 9 days of life (3 – 27); 8 days (3 – 17) in FSG and 19 (6 – 27) in SSG. The dose of L-T4 differed between FSG and SSG at all three analysed time points (start of the therapy, 12 months, and 3 years) with significantly higher doses in FSG. The same was observed for the patients with thyroid dysgenesis vs. GtS. Screening TSH level was $\geq 28\text{mIU/L}$ in 91.7% of patients with thyroid dysgenesis in comparison to 74.0% of patients with GtS ($p = 0.038$). The optimally treated group (fT4 in the upper half of the reference range, according to the guidelines) was up to 58.0% of the children during the follow-up. The risk for overtreatment was present in 1/5 of the study group after 12 months and 1/4 after 3 years of L-T4 therapy. Analysis of new TSH thresholds showed an increased prevalence of mild hypothyroidism, GtS, and either euthyroid state or overtreatment while treating with lower L-T4 doses in comparison to the

Research In Congenital Hypothyroidism

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