FREQUENCY OF COMMON CAUSES FOR CONGENITAL HYPOTHYROIDISM USING THYROID SCINTIGRAPHY: THE HOSPITAL FOR SICK CHILDREN EXPERIENCE

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Background/Purpose: Neonatal screening TSH and T4 tests are used for initial diagnosis of congenital hypothyroidism (CH) and TC99m-thyroid scan (TS) is suggested for further investigation to understand the possible etiology. Determining frequency of each underlying causes of CH is important for evaluation of disease severity, outcome and treatment regimens. The purpose was to determine the frequency for common causes of CH with TC99m thyroid scan from a large sample size.

Methods: 477 newborn patients with elevated TSH level (>20 IU) on screening tests and diagnosis of CH who referred for the TS between January 2004 and March 2013 were retrospectively reviewed. Based on TS, the thyroid gland was classified as normal scan, ectopic (lingual or sublingual), increased uptake (including dyshormonogenesis) and decreased uptake (including agenesis and hemiagenesis).

Results: The average age was 0.09, with 210 male and 267 female patients. A total of 112/477 (23%) were found to have an ectopic gland, with 33% males and 67% females. 159/477 (33%) demonstrated reduced thyroid tissue or hemiagenesis, with 50% males and 50% females. 133/477 (28%) of the scans demonstrated increased uptake or dyshormonogenesis, with a 43% to 57 % male to female distribution. Finally, 73/477 (15%) patients were found to be normal, 58% male and 42% female.

Discussion & Conclusion: Tc-99m Thyroid Scintigraphy is useful for the initial investigation of suspected CH and identifying underlying pathology. Ectopic gland (more common in females patients), reduced thyroid tissue uptake or hemiagenesis, and increased thyroid uptake including dyshormonogenesis are the three most common causes of CH.