Hypothyroid Screening in Children with Down Syndrome – A Service Evaluation

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Background & Aims

Individuals with Down Syndrome are at an increased risk of developing thyroid disease[1]. Given that thyroid disorders represent a preventable cause of neurodevelopmental impairment[2], early detection and treatment are essential to maximise cognitive abilities. This service evaluation sought to assess the efficacy of the Down Syndrome Hypothyroid Screening Programme in its uptake and subsequent diagnosis of hypothyroidism.

Methods

A report of all children with known Down Syndrome was obtained from the Greater Glasgow and Clyde Down Syndrome database. Children were excluded if they were under the age of 2 years or had been a resident for less than 2 years within the region. Electronic Patient Records were used to access baseline characteristics and results of venous Thyroid Function Tests (TFTs) for all children born with Down Syndrome. Data on TSH capillary screening were obtained via the Scottish Newborn Bloodspot Screening lab. Data were collected on each child's previous 3 screening samples and the time between screening was calculated in decimal years. From this, the children referred to Endocrinology following abnormal screening and those subsequently commenced on levothyroxine therapy following referral were identified.

Results

We identified 248 children with Down Syndrome (122 male). 20 children were excluded as they were <2 years old or residents for <1 year. This left 228 children (114 male). The mean age of our cohort was 9.9 years, range (2.1-22.7). 3 children received no screening in their lifetime.

1. Prasher V. *Down Syndrome and Thyroid Disorders: A Review.* Down Syndrome Research and Practice. 1999;6(1), 25-42.
2. Prezioso G, Giannini C, Chiarelli F: Effect of Thyroid Hormones on Neurons and Neurodevelopment. Horm Res Paediatr 2018;90:73-81.

As can be seen in Figure 1, of those screened, 92% of patients received screening within the last 1.0 decimal years (207/225) and a further 3.1% (7/225) of patients received screening within the last 1.5 years. 7 of the 225 children had only been screened once (n=218). 74 children (33.9%) had 1.0 years or less between their previous screenings. A further 118 children (54.1%) had 1.5 years or less between each screen. This can be seen in Figure 2.

1

Time since previous screening (years)	Number of patients (%)	
0-0.5	65 (28.9)	
0.5-1.0	142 (63.1)	95.1%
1.0-1.5	7 (3.1)	
1.5-2.0	7 (3.1)	
2.0-2.5	0 (0.0)	
2.5-3.0	2 (0.9)	
3.0-5.0	0 (0.0)	
>5	2 (0.9)	

2

Time between screening (years)	Number of patients (%)	
0-0.5	9 (4.1)	
0.5-1.0	65 (29.8)	88.0%
1.0-1.5	118 (54.1)	
1.5-2.0	14 (6.4)	
2.0-2.5	6 (2.8)	
2.5-3.0	4 (1.8)	
3.0-5.0	2 (0.9)	
>5	0 (0.0)	

1- Time in decimal years from final date of data collection (5/12/2019) to previous screening
2- Time in decimal years between previous screening and screening immediately prior

21 children (9.3%) had abnormal screening, which resulted in referral to Endocrinology for 20 children. One child had normal TFTs and so was not referred. Of the 21 children with abnormal screening, 16 (76.2%) were commenced on Levothyroxine therapy.

Conclusion

Within Greater Glasgow & Clyde, the Hypothyroid Screening Programme is effective in monitoring and detecting thyroid disease. The majority of children with Down Syndrome receive hypothyroid screening annually. Of those screened, 9.3% of children had abnormal screening results, with 76.2% of these children commenced on Levothyroxine therapy.