An Audit of the Management of Thyroid Disease in Children with Down Syndrome

Abstract:
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Abstract

Children with Down syndrome are at a higher risk of thyroid dysfunction than children in the general population. The aim of this audit was to determine thyroid screening practice at University Hospital Limerick and to compare it to the Irish guidelines for the management of children with Down syndrome. The thyroid function tests (TFTs) of 148 children with Down syndrome were assessed through retrospective database review. Overall compliance with the guidelines was 79/148 (53%), although this varied by age category. The 0–5 years category had a compliance rate of 47/54 (87%), the 6–11 years category was 22/51 (43%), and the 12–17 years category had a compliance rate of 10/43 (23%). The guidelines are effective for monitoring purposes, although performing an annual TFT throughout childhood may be warranted.

Introduction

Thyroid dysfunction is common in children with Down syndrome. Subclinical hypothyroidism i.e. an elevated thyroid stimulating hormone (TSH) result in the presence of normal thyroid (T4) and triiodothyronine (T3) is particularly common in children with Down syndrome with prevalence ranging from 25.3% to 50%.[6] The Irish guidelines for screening for childhood thyroid dysfunction in children with Down syndrome were last reviewed in 2009 and recommend routine TSH after birth (on Guthrie screening test), then TSH and free T4 annually until age 5 years; and TSH and free T4 every 2 years thereafter. Thus, every child with Down syndrome should have had at least 1 TFT performed in the past 2 years. A retrospective database review was carried out with the purpose of auditing clinical practice in the University Hospital Limerick Complex (UHL Complex). The aim of this audit was to determine thyroid screening practice at UHL and to compare it to the Irish guidelines for the medical management of children with Down syndrome.

Methods

The study design was an audit which was conducted through a retrospective database review. Children with karyotype confirmed Down syndrome aged 0-18 years were eligible for inclusion in this study. There were no exclusion criteria. Searching local hospital (community and clinic) databases identified 148 children with Down syndrome attending services in Limerick, and all were included in this audit. The patients were out-patients of the UHL or attended a community clinic. The patients attended Early Intervention Clinics/School Age Disability Team Clinics, HSE Mid West Regional Disability Services and General Paediatrics Clinics at UHL. None attended Paediatric Endocrinology. All were under the management of a Paediatrician at UHL. The cohort was sub-divided by age as follows: 54 patients in the 0-5 years category, 51 in the 6-11 years category, and 43 in the 12-17 years category.

All TFTs were venous samples analysed in the UHL central laboratory. Plasma FT4 was measured by IMMULITE 2000 Free T4, a solid-phase, enzyme-labelled chemiluminescent competitive immunoassay. The reportable range of this assay is 3.9–77.2 pmol/L. TSH was measured by IMMULITE 2000 Rapid TSH, a solid-phase, chemiluminescent immunometric assay. The assay range of this assay extends to 75mIU/ml. Current cost of an FT4 test in the UHL central laboratory is €1. A TSH assay also costs €1. The results of all TFTs (TSH, free T4, T4 and thyroid autoantibodies) for each child with Down syndrome were gathered and which could be expected to influence the frequency of testing includes: the presence of autoimmune thyroid disease, presence of thyroid autoantibodies, and history of thyroid dysfunction in first degree relatives. Therefore, the results of children with a diagnosed thyroid disorder and consequent increased monitoring, will not change the frequency of repeat TFTs performed. The clinical information which was not gathered and which could be expected to increase the frequency of testing includes: the presence of autoimmune disease; cardiac disease; or thyroxine use in the cohort. We anticipate that a prospective study will address these limitations. Also each child’s test results were interpreted according to recommendations for their specific age; therefore the results of children with a diagnosed thyroid disorder and consequent increased monitoring, will not significantly affect the number of abnormal test results in this study.

Results of the Audit

Overall compliance with the guidelines was 53% (79/148) since 2001. Compliance is highest at 87% (47/54) in the patient age category 0–5 years old. Compliance in the patient age category 6–11 years old is 43% (22/51). The lowest compliance is for the patient age category 12–17 years at 23% (10/43) (see Figure 2). Of the 148 patients, 117 (79%) have had a TFT in the last two years, which is the minimum time recommended for a TFT check, regardless of the age of the child. Thus, 31 (21%) patients had not received a TFT in >2 years.

Discussion

Although overall compliance with the guidelines since 2001 is just 53%, the percentage of patients who have received a TFT in the last 2 years is 79%. This suggests a recent improvement in compliance. Overall, just 23% of patients have gone >2 years since having received a TFT. This study has limitations; it is a small, retrospective study. It is possible that children that had TFTs performed that were not captured in our records. If this is the case, our results underestimated compliance. This is disappointing, given that this study suggests non-compliance in 1 in 5 children in the past 2 years. Also, our databases are limited and do not allow for an accurate review of the clinical details of children diagnosed with hypothyroidism. The clinical information averages for TSH and free T4 and also change the frequency with which these children have repeat TFTs performed. The clinical information which was not gathered and which could be expected to increase the frequency of testing includes: the presence of autoimmune disease; cardiac disease; or thyroxine use in the cohort. We anticipate that a prospective study will address these limitations. Also each child’s test results were interpreted according to recommendations for their specific age; therefore the results of children with a diagnosed thyroid disorder and consequent increased monitoring, will not significantly affect the number of abnormal test results in this study.

If the Irish guidelines are followed consistently, the expectation would be a reduction in the number of children with Down syndrome presenting with overt hypothyroidism. However, the risk of acquiring hypothyroidism increases with age in children with Down syndrome. Thus, the Irish guidelines may need to introduce yearly thyroid screening after 5 years of age. Other national guidelines suggest more frequent thyroid screening in this population. More evidence is needed regarding the point at which treatment should be commenced in cases of subclinical hypothyroidism. Children with Down syndrome are at risk of developing health issues and attend many medical appointments. Any move to increase this burden should be carefully considered. Furthermore, more appointments and tests imply increased healthcare costs.

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The live birth rate in Ireland in 2010 was 73,724. The estimated prevalence of Down syndrome in Ireland is 1 in 546 live births. Thus in 2010, the estimated number of babies born with Down syndrome was 135. The increased cost of 8 extra TFTs for these children up to age 18 years is €6480; including the cost of the assays only. The cost-benefit of this guideline change should be analysed scientifically after any change in practice.

In conclusion, compliance with the guidelines is highest in the 0-5 years age group, and lowest in the 12-17 years age-group. Consideration should be given to amending Irish guidelines to include TFT annually throughout childhood as well as offering guidelines on the management of subclinical hypothyroidism in this population. Prospective Irish studies are required to evaluate the benefits and disease prevention achieved by our current guidelines and any further benefits that may be realised by changing current Irish guidelines.

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