Down Syndrome Medical Interest Group

1736 NEWBORN SCREENING AND SURVEILLANCE OF THYROID DISORDER IN INFANTS WITH DOWN SYNDROME

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Background The Down Syndrome Medical Interest Group U.K. & Ireland published guidelines on thyroid disorders in children and young people with Down syndrome: surveillance and when to initiate treatment in April 2020.

The guidelines recommend that infants with Down syndrome be offered an initial blood spot in the neonatal period in accordance with the current national newborn screening programme for congenital hypothyroidism.

The guidance also recommends that all infants with Down syndrome are offered thyroid function testing at 4–6 months of age and that no additional testing is required in the neonatal period unless thyroid dysfunction is suspected or where additional testing is recommended by the national newborn screening programme.

Objectives To ascertain whether clinicians have adhered to the new guidance on offering thyroid function testing as per the Down Syndrome Medical Interest Group guidance.

Methods A retrospective notes review was undertaken of all infants with Down syndrome referred to the three child development centres in Leeds during the period February 2020 – February 2021.

Results Electronic case notes of 13 babies with Down Syndrome were reviewed. Six infants were offered thyroid function testing between 4–6 months of life. Although two babies had insufficient samples and so did not get a result. At the time of auditing these babies were 26 weeks and 49 weeks old and yet to have repeat blood sampling.

One infant was tested at 7 months and 1 infant was tested at 8 months.

Three infants are yet to have thyroid function tests and are more than six months of life. One infant is only three weeks old and too young to be offered testing but this has been arranged to be taken at 4 months of life.

One infant (male) had thyroid function testing at 21 days of life due to abnormal TSH on newborn screening and has subsequently been diagnosed with congenital hypothyroidism.

One infant had inappropriate thyroid function tests sent at 10 days of life whilst on the neonatal unit.

One baby had a TSH of 6.4 with a normal T4 and the decision was made to repeat the samples in 3 months' time and not within 1–5 days as per recommended guidance.

Four babies had blood spot TSH (including the two babies with insufficient samples) and 4 babies had venous samples sent.

All babies were offered newborn blood spot screening.

The parents of ten babies were informed on thyroid dysfunction in Down syndrome and the importance of monitoring thyroid function in the initial clinic by the clinician. For the remaining 3 babies there is no documented evidence that this information was conveyed.

Conclusions We can do better in terms of timely offering the initial surveillance testing at 4–6 months of life and following up promptly on arranging repeat testing if there are sampling issues.

We must continue to educate parents on thyroid dysfunction in Down syndrome ensuring they are aware of key signs and symptoms.

Association of Paediatric Emergency Medicine

1739 BPSU STUDY ON SEVERE ACCIDENTAL POISONING IN CHILDREN (ASPIC)

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Background Accidental poisoning in children is a common reason to attend hospital. Children, particularly under 5 years, are naturally inquisitive and taste and swallow products, some of which may be harmful. A small select number of medications and chemicals are subjected to child resistant packaging legislation which has reduced harm to children. However most adult medicines are not, and one or two doses of these could prove fatal to a toddler. Additionally, teenagers may engage in illicit drug or alcohol use resulting in unintentionally poisoning themselves as a result of risk-taking behaviour.

Methods Thirteen months surveillance, where Consultant Paediatricians within UK/ROI reported cases of severe accidental poisoning which resulted in death and/or signs/symptoms requiring significant interventions in children < 15 years via the BPSU Orange Card. Further history, clinical details, and management were requested via a follow up questionnaire. Poisonings were classified using The European Association of Poisons Centres and Clinical Toxicologists (EAPCCT) poisoning severity score (PSS), which looks at the clinical symptoms and management of a poisoning episode (see table). Cases with a PSS < 2 were excluded from the analysis.

Results Of 116 cases, 30 were lost to follow-up, 13 were excluded, 31 reported in error, 3 were duplicate cases and 7 did not meet the case definition. This left thirty-two cases that met the surveillance case definition, and one was removed as it did not meet the analytic case definition of a PSS ≥ 2.

71% cases involved children under 5 years with 39% less than 2 years and 68% were male. 84% of cases occurred in either the child's own home (16/31), or a family member (10/31). 13/31 involved a prescribed/over-the-counter medication which were contained in blister packs (5/31), a bottle/jar (2/31), or loose (2/31). Commonly, these were opiates, sedatives or psychiatric medications. 12/31 cases involved illicit substances or alcohol.

Abstract 1739 Table 1

Poisoning Severity Score (PSS)	Cases
NONE (0): No symptoms or signs	
MINOR (1): Mild, transient and spontaneously resolving symptoms	
MODERATE (2): Pronounced/prolonged symptoms	14
SEVERE (3): Severe/life-threatening symptoms	16
FATAL (4): Death	1

The commonest symptoms were reduced GCS (20/31), respiratory depression (11/31), seizures (10/31) and tachycardia (8/31). Most commonly, cases required 3–5 different managements. These were: continuous oximetry PLUS oxygen PLUS ECG monitoring (68%), GCS <12 AND frequent GCS monitoring (68%), cardiovascular monitoring (65%), and invasive ventilation (48%).

Conclusions Despite relatively small numbers, our study shows that accidental poisoning is a preventable condition. Significant poisoning can cause serious symptoms requiring various treatments, but rarely results in death. Our study demonstrates that adult medications that are currently not subject to child resistant packaging laws are causing significant harm to children. This highlights that further legislation is necessary on all medications to prevent harm and even death in children. In all ages, a large proportion of episodes involved illicit drug use suggesting further public health medicine and drug safety campaigns are required to educate both young people and also families around the risks and dangers of drug use.

Down Syndrome Medical Interest Group

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PROFILE OF THYROID DISORDERS IN CHILDREN WITH DOWN SYNDROME

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Background The prevalence of thyroid disorders in children with Down syndrome is 6–10%. Guidelines on thyroid disorders in children and young people with Down syndrome: surveillance and when to initiate treatment in April 2020 were published by the Down Syndrome Medical Interest Group U.K & Ireland in 2020. The spectrum of thyroid disorders in children with Down syndrome includes congenital hypothyroidism and autoimmune thyroid disorders.

Objectives We undertook a retrospective study to review the profile of thyroid disorders of children with Down syndrome currently served by our child development centre serving a child population (0–19 years) of 64,500 children.

Methods A retrospective electronic case notes review was undertaken of 69 children with Down syndrome attending the child development centre, to identify children with a diagnosis of thyroid disorder and assess their biochemical and clinical presentation

Results Electronic case notes of 69 children with Down syndrome were reviewed. One infant (male) was diagnosed with congenital hypothyroidism. Six children were diagnosed with autoimmune hypothyroidism. Prevalence rate 8.8 percent. The median age at diagnosis was 6.6 years. The gender ratio was 3 female: 4 male. Two of these children had a borderline TSH for prolonged period before formally receiving a diagnosis of hypothyroidism, this ranged between 8 months and 45 months, the thyroid function was monitored every 6–12 months during this period. At the time of the initial raised TSH levels the TPO antibodies were normal and increasing to 997 and >1300. Two children have free T4 levels above the normal range (21) despite their TSH levels being above the local reference range and good compliance with medication. There was a rise in BMI at the time of diagnosis in six children (data not available for remaining children). Symptoms noted at diagnosis of thyroid disorder were weight gain,

tiredness and sleep disturbance particularly in female patients. None of the children were recorded to have goitre. Two additional children were noted to have persistently raised TSH levels currently undergoing close monitoring, interestingly both these children have a slight rise in their TPO levels but less than 100 and a marginal increase in their BMI at the time of the initial rise in TSH levels, both sets of parents declined repeat serum thyroid testing within 1–5 days as recommended in the updated guidelines. There were no children diagnosed with hyperthyroidism or Graves' disease.

Conclusions Thyroid disorders in children who have Down syndrome appear to follow a more insidious course with borderline or subclinical hypothyroidism being more commonly present than the general population. It is important to closely monitor the thyroid function to prevent additional disability. With the introduction of earlier thyroid surveillance at 4–6 months as per the updated DSMIG guidelines, it is important to undertake large-scale prospective population studies to evaluate the developmental outcomes in children with Down syndrome and subclinical hypothyroidism.

British Paediatric Neurology Association

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IT'S NOT ALWAYS ABOUT HONEY

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Background Infantile Botulism (IB) is a rare, but potentially fatal disorder typified by flaccid paralysis in infants. It is caused by toxins released by gram-positive anaerobic bacteria, *Clostridium botulinum* - soil organisms that exist as spores. These botulinum spores are consumed (directly or through contaminated foods such as honey or home-canned foods) and they colonise the large intestine of infants before releasing the botulinum toxins that bind irreversibly to the neuromuscular junction causing flaccid paralysis. Paralysis especially of the muscles of respiration place the affected infant at risk of death if respiratory support is not given

Objectives To review a case of IB and its management.

Methods Case report.

Results A 6-month-old male infant with no significant past medical, family, and birth history, presented to the Children's Assessment Unit with cough, decreased feeding, and lethargy at the outset of neurologic symptoms, and respiratory distress. Further anamnesis revealed no sick contacts, honey ingestion, recent travels, and indoor pet exposure. His review of systems was negative.

Upon initial assessment the infant's vital signs were normal and there were no notable findings on systemic examination apart from bilateral crackles and wheeze. However, his respiratory status worsened in less than 24 hours after admission, resulting in his mechanical ventilation and PICU admission.

The patient's PICU course was notable for the development of a broad differential consequent of his unexplainable and deteriorating clinical status. The diagnosis of infantile botulism was eventually entertained on the heels of an unremarkable extensive lab and imaging testing. POSITIVE Murine micro-assay for botulinum toxin was confirmatory.

The infant was treated with botulinum immunoglobulin (Baby BIG) on day 13 of his admission with remarkable