Down Syndrome Medical Interest Group

A RETROSPECTIVE STUDY OF COELIAC DISEASE SCREENING AMONG CHILDREN AND YOUNG PEOPLE WITH DOWN’S SYNDROME

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Background Coeliac disease (CD) is an immune-mediated condition which develops in genetically susceptible individuals and is triggered by the ingestion of gluten. An estimated 1% of the UK population have CD. In persons with Trisomy 21 or Down’s syndrome (DS), the lifetime prevalence of CD is significantly increased (5–12%). National guidance therefore recommends routine screening for CD among children and young people (CYP) with DS. This specifies an initial screen of HLA-DQ status, which need only be followed by subsequent tTG-IgA surveillance in HLA-DQ2 or HLA-DQ8 positive subjects.

Objectives We sought to determine the prevalence of CD among CYP with DS in our region and describe current screening practices in this at-risk group.

Methods Data were collected retrospectively from an active database of CYP diagnosed with DS being managed by the Leicester and Leicestershire Community Paediatrics service. Subjects were excluded if younger than six months or where electronic records were unavailable. Baseline characteristics and data on timing, frequency and tests used for coeliac screening were collected from electronic records and analysed using Microsoft Excel.

Results We identified 257 children with DS under active follow-up with the Leicester and Leicestershire Community Paediatrics service. Four were excluded (3 had no laboratory records and 1 was <6 months). Among 253 included children, there was a male preponderance (58% male) with age at time of survey ranging from 6 months to 21 years, mean (SD) 8.7 (4.8) years. At least one tTG-IgA result was available in 147 (58%) children. HLA typing had been performed in 9 (3.6%) children (8 with confirmed CD). Overall, CD was confirmed in 14 (5.5%) children. The mean age (SD) at diagnosis was 7.5 (3.8) years, 12 (85.7%) were symptomatic pre-diagnosis and 9 (64.3%) had tTG-IgA >128. Anti-endomysial antibodies were positive in 12/14 (85.7%) children. Overall, 106 (42%) children had neither a tTG-IgA nor HLA typing performed. Among the children without CD with available tTG-IgA results, 61/133 (45.9%) had a tTG-IgA result within the last 15 months. The mean (SD) frequency of tTG-IgA testing in those with at least one tTG-IgA was every 4.0 (4.8) years. Limitations to this study include the retrospective design.

Conclusions In this local cohort of 253 CYP with DS, CD prevalence was 5.5% in keeping with national trends. However, routine tTG-IgA surveillance was less frequent than is recommended, which risks missing detection of asymptomatic patients. The majority of CD confirmed cases were symptomatic prior to their diagnosis. Improved screening and earlier diagnosis of CD among CYP with DS may therefore help to reduce morbidity. In this community setting, testing of HLA-DQ status was not routinely practiced. Early knowledge of negative HLA-DQ2/8 status can reassure most parents that their children do not have a CD risk. This negates the need for serial tTG-IgA testing in some CYP and is a more cost-effective strategy. We intend to disseminate these results locally with re-audit in two year’s time.

Quality Improvement and Patient Safety

COVID-19, LOCKDOWN 1.0, AND THE MOVE TO TELEMEDICINE: IMPACT ON GLYCATED HAEMOGLOBIN IN PAEDIATRIC DIABETES MELLITUS

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Background Maintaining blood glucose levels within target is the cornerstone of diabetes mellitus management, reducing the risk of complications. Glycated haemoglobin (HbA1c) is the gold standard assessment, reflecting plasma glucose over 2–3 months. NICE guidance recommends four clinic attendances and HbA1c measurements per year, with a target of 48mmol/mol or lower. The National Paediatric Diabetes Audit showed a mean of 64.6mmol/mol in 2018/19. At a UK paediatric diabetes unit, face-to-face clinics (F2F) were converted to telephone appointments on 30/03/2020 due to the first COVID-19 UK lockdown. There was a phased return to F2F and HbA1c testing from June 2020.

Objectives To determine whether the COVID-19 pandemic and consequent national lockdown and move to telemedicine affected HbA1c levels in children with diabetes mellitus.

Methods HbA1c results were recorded throughout 2020, excluding those diagnosed in 2020. Each patient’s final HbA1c in January-March before lockdown (Pre-LD) was compared to both their first HbA1c after lockdown (Post-LD) and the mean of all of their HbA1c’s after lockdown (Av-Post-LD). Comparisons were analysed grouping patients by Pre-LD, which was assumed to be their baseline.

Results Of the 258 patients, 61 (23.6%) had no Pre-LD and 38 (14.7%) had no Post-LD, excluding them from further analysis. Numbers of F2F and HbA1c testing varied throughout the year; 92 tests were done in January, pre-pandemic, falling to 1 in April, peaking at 83 in September, dipping to 27 in December. Number of tests per patient post-lockdown varied from 1–5 (mean 1.45). When comparing results grouped by baseline, a correlation was seen (table 1).

Abstract 1269 Table 1

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<th>Group</th>
<th>Number of Subjects</th>
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<th>Post-LD (2)</th>
<th>Av-Post-LD (3)</th>
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Improved 1–2 and 1–3 have a positive correlation (p<0.05).