



Annual Health Check Information for GPs

Gastrointestinal Disorders

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Below is Down's syndrome specific information. The information is for GPs and is to be used alongside DSA's Adult Health Book.

Children and adults with Down's syndrome will exhibit gastrointestinal symptoms from time to time such as vomiting, diarrhoea, constipation, abdominal pain and discomfort that resolve with minimal or no intervention much as in others. However, they may develop structural and functional disorders of the gastrointestinal tract and related structures more commonly. How commonly these occur in the general population of Down's syndrome is unknown because figures have been derived from selected populations such as those attending special clinics for Down's syndrome when about 10% of children and teenagers will be affected ¹. Over three quarters of neonates attending clinics may have gastrointestinal problems including feeding difficulties or developmental anomalies ².

Structural problems may affect the gastrointestinal tract from the mouth to the anus but many conditions will occur in Down's syndrome with similar frequency to other children. However, oesophageal, duodenal, and small bowel atresia or stenosis, annular pancreas causing small bowel obstruction, imperforate anus and Hirschsprung disease may be more common than in the general population ³.

Obstruction in the gastrointestinal tract may be detected before birth by imaging techniques and so allow for planned intervention early after birth. If diagnosis is not made pre-birth no bowel actions, vomiting and a distressed baby indicating abdominal pain will suggest bowel obstruction and the need for urgent surgical intervention. Imperforate anus either total or partial may also occur and require surgery. Hirschsprung disease affects about 2% of those with Down's syndrome and manifests as a distended abdomen, poor weight gain, vomiting and constipation. Short segment disease can be difficult to diagnose.

Gastro-oesophageal reflux should be suspected in a child who appears uncomfortable during or after feeding. Children with Down's syndrome are prone to this because they spend less time in the sitting position and muscle tone in the lower oesophageal sphincter may be reduced thus allowing reflux. It is possible that developmental abnormalities in the enteric nervous system also have a role to play here and also perhaps in other functional disturbances. Too liquid feeds may contribute to the problem. Aspiration pneumonia may be a presenting feature of reflux and early evaluation of oesophageal function should be undertaken in children with

chronic cough or recurrent pneumonia. Reflux can easily be misdiagnosed as asthma and so remain untreated.

Adults with Down's syndrome are also prone to a wide range of gastrointestinal problems including reflux, obesity, constipation and diarrhoea. Infection with H.pylori appears to be more common but the implications are not clear. Non-immunity to hepatitis A and B can be high and indicates the need for immunisation.

Coeliac disease (CD) that is associated with Down's syndrome can present at any age. Symptoms in children and adults are protean and include growth failure, malaise, vomiting, abdominal distension, diarrhoea and constipation. Unexplained anaemia, iron and calcium deficiency, point to the diagnosis.

Screening studies have shown a prevalence of CD in Down's syndrome of about 5% and because of this strong association some have advocated screening all subjects using anti-tissue trans-glutaminase and/or endomyseal antibodies ⁴. Screening should begin at the age of 3 years and be repeated every 2-3 years since a single negative test will not rule out CD for life. By establishing the HLA status of individuals and excluding those from the programme who do not carry HLA-DQ2 or HLA-DQ8, markers that are necessary for CD to develop, the number of screening tests can be reduced by 60%. Screening has the potential to diagnose all cases irrespective of symptoms. Whether this is an effective approach is still not clear because those with minimal or no symptoms may not be persuaded to undergo a small bowel biopsy to confirm the diagnosis or adhere to a gluten free diet.

A second approach to make the diagnosis of CD in Down's syndrome is a case-find strategy that targets only those with clinical features consistent with the diagnosis e.g. symptoms, unexplained anaemia, family history. If the diagnosis is suspected, CD specific antibodies should be looked for and if positive a small bowel biopsy advised to confirm the diagnosis. Antibody negative CD occurs and may be due to a false negative test or IgA deficiency in which case IgG based tests are available. If the diagnosis of CD is strongly suspected a duodenal biopsy should be advised even in the absence of antibodies.

Some patients with high levels of tTG (>10 times the upper limit of normal) may not require biopsy to establish the diagnosis of CD because this accompanies diagnostic histology. This has already been acknowledged in paediatric guidelines ⁵ and it is likely to be applied soon to adult practice. Not having to biopsy some with Down's syndrome would be advantageous.

If CD is diagnosed a gluten free diet should be offered and the opportunity to see a dietician experienced in managing the diet. Carers and where possible patients should be involved at all stages of the diagnostic and management process.

References

1. Yam, W.K.-L., Tse, P.W.T., Yu, C.M., Chow, C.B., But, W.M., Li, K.Y., et al. Medical issues among children and teenagers with Down syndrome. *Down Syndrome Research and Practice* 2008;12(2):138-40.
 2. Spahis, J.K. & Wilson, G.N. Down syndrome: Perinatal complications and counseling experiences in 216 patients. *Am J Med Genet* 1999;89(2):96-9.
 3. Buchin, P.J., Levy, J.S. & Schullinger, J.N. Down's syndrome and the gastrointestinal tract. *J Clin Gastroenterol* 1986;8(2):111-4.
 4. Marild, K., Stephansson, O., Grahnquist, L., Cnattingius, S., Soderman, G. & Ludvigsson, J.F. Down syndrome is associated with elevated risk of celiac disease: a nationwide case-control study. *J Pediatr* 2013;163(1):237-42.
 5. Husby, S., Koletzko, S., Korponay-Szabo, I.R., Mearin, M.L., Phillips, A., Shamir, R., et al. European Society for Pediatric Gastroenterology, Hepatology, and Nutrition guidelines for the diagnosis of coeliac disease. *J Pediatr Gastroenterol Nutr* 2012;54(1):136-60.
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The Down's Syndrome Association (DSA) is the only organisation in England, Wales and Northern Ireland which supports people with Down's syndrome at every stage of life.

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