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| **Coeliac disease** |
| **­­**  **Definition of coeliac disease:**   * An autoimmune enteropathy of the small bowel characterised by mucosal inflammation on gluten exposure leading to malabsorption. |
| **Epidemiology of coeliac disease:**   * Globally, the prevalence is 1%. However, rates vary from as little as 1 in 300 to 1 in 40. * In the UK, the prevalence is 0.5-1% * Can occur at any age but peak incidences are in infancy and 5th decade. * It rarely affects individuals of African, Chinese or Japanese descent. * Women affected more than men – approx. 2:1 |
| **Aetiology of coeliac disease:**   * T-cell mediated autoimmune response to dietary prolamin exposure (proteins in wheat, rye, barley etc such as gliadin) giving an inflammatory response causing small bowel villous atrophy and thereby leading to malabsorption. * Genetic factors (10-15% first degree relative concordance, 70% identical twin concordance)   + HLA-DQ2 (Sens 94%, spec 73%)   + HLA-DQ8 (Sens 12%, spec 80%) * Other factors   + Possible early viral trigger – potentially rotavirus or intestinal adenovirus   + Some evidence suggests exposure to grains in first 3 months of life increases risk   + Prolonged breast-feeding may reduce the risk of childhood coeliac * Association with other autoimmune conditions such as type 1 diabetes, primary biliary cirrhosis and autoimmune thyroid disease (both hyper and hypothyroidism). |
| **Presentations of coeliac disease:**   * History * Asymptomatic (up to 1/3 cases) * Fatigue and malaise * Abdominal pain and bloating * Diarrhoea * Steatorrhoea * Nausea/vomiting * Weight loss and failure to thrive (children especially) * Lactose intolerance (can develop secondarily with chronic disease) * Subfertility and recurrent miscarriages * Examination   + Anaemia   + Angular stomatitis   + Mouth ulcers   + Dermatitis herpetiformis     - Blistering subdermal eruption associated with coeliac disease, usually very itchy |

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| **Differential diagnosis of coeliac disease:**   * Irritable bowel syndrome (IBS) * Inflammatory bowel disease (IBD) * Chronic infective gastroenteritis e.g. Giardiasis * Lactose intolerance * HIV enteropathy * Bacterial overgrowth |
| **Investigations and diagnosis of coeliac disease:**   * Blood tests   + Full blood count: anaemia – can be microcytic due to iron deficiency; macrocytic due to folate and/or vitamin B12 malabsorption; or a mixed picture.   + Haematinics (i.e. folate, vitamin B12, ferritin, total iron levels)   + Low calcium and vitamin D levels with compensatory secondary hyperparathyroidism   + Liver function tests – these can be abnormal and thought to be due to a non-specific hepatitis * Anti-endomysial and anti- tissue transglutaminase antibodies (TTG Ab)   + Both very sensitive and specific (sens 99%, spec >90%)   + Do TTG (IgA) first as it is simpler, if equivocal result then do antiendomysial (IgA)     - 10% coeliacs are IgA deficient so check this as well * Duodenal/jejunal biopsy – gold standard of diagnosis   + Duodenal biopsy as good as jejunal as long as ≥ 4 biopsies taken   + Subtotal villous atrophy; crypt hyperplasia; intra-epithelial lymphocytosis   + Modified Marsh criteria     - Five stages from 0 (normal mucosa) to 3c (crypt hyperplasia and total villous atrophy) * DEXA scanning   + Should be done in all patients as calcium and vitamin D malabsorption can lead to osteopenia |
| **Management of coeliac disease:**   * Total adherence to gluten-free diet   + Found in wheat, barley and rye and oats   + Symptoms and microscopic gut changes should resolve with gluten-free diet   + Dietician involvement is usually very helpful as are patient support groups and networks * Iron, calcium, folate and vitamin B12 supplementation * Dapsone 50-100mg once daily can improve dermatitis herpetiformis although this will also improve with a gluten-free diet. * 5-yearly pneumococcal vaccination is recommended due to hyposplenism |

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| **Complications of coeliac disease:**   * Failure to improve despite gluten-free diet   + Only 10% of those who report non-responsive disease will have true refractory coeliac disease.   + Check compliance. Re-test antibodies if compliance is unsure. * Enteropathy-Associated T-Cell Lymphoma (EATCL) * The risk of all gastrointestinal malignancies, and in particular oesophageal, is slightly increased (but incidence decreased by gluten-free diet) * Dermatitis herpetiformis * Osteopenia and osteoporosis * Hyposplenism   + Occurs in approx. 1/3 cases. Can predispose to infection (especially encapsulated bacteria) * Lactose intolerance * Bacterial overgrowth * Pancreatic insufficiency |
| **Prognosis of coeliac disease:**   * Recent population-based studies have shown only a minor or a non-significant increase in mortality in patients with coeliac disease * Severity of disease at presentation and poor adherence to a gluten-free diet are predictors of increased mortality. |
| **Common questions concerning coeliac disease:**   * What are the microscopic features on duodenal biopsy?   + Crypt hyperplasia; villous atrophy; epithelial lymphocytosis * What are the common complications of coeliac disease?   + Iron-deficiency anaemia; folate and vitamin B12 deficiencies; osteoporosis |