Jejunal Discaccharidases

Pseudonyms: Jejunal biopsy analysis; sucrase, lactase and maltase.

Disaccharidase enzymes are found only in the mucosa of the duodenum and jejunum. They hydrolyse disaccharides to monosaccharides. The method can be used to distinguish between different enzymes hydrolysing lactose in the small intestine, viz. brush border lactase (lactase proper) which is absent or reduced in alactasia (lactose intolerance) and coeliac disease, and acid $^{\beta}$-galactosidase, an intracellular enzyme which persists in these disorders.

Presenting symptoms may include abdominal pain, flatulence, diarrhoea and nausea. The strength and frequency of these is related to the amount and type of food recently consumed, the amount of the disaccharides they contain and the degree of enzyme deficiency. The literature suggests biopsy should only be carried out when symptoms have been present for at least 3 weeks with coeliac disease and infective causes excluded.

General Information

Collection container: Biopsy samples should arrive on ice in a plain Universal tube without any liquid, as soon as possible after collection. Tissue must be received in the laboratory within 1 (one) hour of collection. Please keep as cool as possible whilst transporting the specimen, preferably using liquid nitrogen or ice. Using the porters to transport the specimen is not recommended.

Contact the laboratory on (70) 11215 (Specialist assay lab) or (27) 64699 (Specialist Biochemistry Reception)

Type and volume of sample: A tiny mass can be analysed (minimum 1mg wet weight) but larger samples will give better estimates of enzyme activity. Please state whether the biopsy has been collected from the jejunum or duodenum as this affects interpretation (see factors known to affect the result below).

Specimen transport/special precautions: Patients should stay on a normal diet (including gluten) for 2 weeks prior to investigation. Specific fasting requirements prior to anaesthesia will be provided by the clinicians.

External users: Samples received from external hospitals must arrive on dry ice. This is a non-CE marked assay and is not accredited under UKAS ISO 15189. Results should be interpreted with caution.

Laboratory information

Method principle: The glucose liberated from lactose, sucrose and maltose is quantitated using a glucose oxidase reaction followed by reaction of the hydrogen peroxide product with 4-hydroxyphenylacetic acid (4-HPAA), which is oxidised to a highly fluorescent compound.

Biological reference range or cut off:

JEJUNAL MALTASE	130 – 456 U/ g protein
JEJUNAL SUCRASE	30 - 152 U/ g protein
JEJUNAL LACTASE	6 - 55 U/ g protein

Turnaround times: 2 weeks

Clinical information

Factors known to significantly affect the results: Duodenal samples will have lower enzyme activities than jejunal. Delayed transport to the lab will result in lower enzyme activities.

Clinical decision points:

The deficiency may be primary (congenital, genetic) or secondary (due to villus atrophy or destruction) caused by infection, autoimmune disease or iatrogenic).

If only one enzyme activity is reduced, this strongly supports a rare congenital deficiency. A reduced activity for more than 1 enzyme more likely indicates a secondary deficiency but this does not exclude a primary deficiency masked by the secondary cause.

The North European population normally have persistence of lactase, so symptomatic presentation at an early age should be investigated, whereas the Asian population have non-persistance as the norm and frequently do not require investigation. Supplementary investigations may include faecal reducing substances and chromatography and breath hydrogen testing.

Specific inherited defects of lactase or sucrase/maltase activity can occur. Sucrase-Isomaltase Deficiency is extremely rare in the UK. A reduction in all disaccharidase enzymes is seen in coeliac disease and in post infective states, due to loss of villi and reduced mucosal area. Full spontaneous resolution usually occurs in 6 to 12 months.

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