Introduction

Congenital Tufting Enteropathy (CTE; OMIM #613217) is a rare, neonatal-onset autosomal recessive condition characterised by chronic intractable diarrhoea leading to severe malabsorption and significant morbidity and mortality. The severity of intestinal malabsorption in most cases results in complete dependence on long-term parenteral nutrition.

Histologically, affected individuals display focal epithelial tufts in the duodenum and jejunum composed of closely packed enterocytes.

The prevalence of CTE is estimated between 1/50,000 to 1/100,000 in Western Europe but is higher in consanguineous populations and patients of Arabic ethnic origin.

Congenital Tufting Enteropathy is caused by mutations in the EPCAM gene at 2p21, with a founder EPCAM mutation reported in several families of Arabic ethnic origin.

The EPCAM gene (NM_002354.2, ENST00000263735) consists of 9 coding exons and encodes an epithelial cell adhesion molecule.

Referrals

Referrals are accepted from Consultant Clinical Geneticists and Consultant Paediatric Gastroenterologists in the following:

- Patients with clinically suspected Congenital Tufting Enteropathy
- Carrier testing in family members for known familial mutations

For clinical enquiries, please contact Dr Neil Shah, Gastroenterology, GOSH
Tel: +44 (0) 20 7405 9200 ext 5949, email: Neil.Shah@gosh.nhs.uk

Prenatal testing

Prenatal testing is available for families in whom specific mutations have been identified or in whom appropriate family studies have been undertaken - please contact the laboratory to discuss.

Service offered

Analysis for point mutations and small insertions / deletions is by direct Sanger sequencing of the coding region in 9 amplicons.

Technical

Mutation screening is carried out by direct sequencing analysis.

Target reporting time

The target reporting time is 8 weeks for an EPCAM mutation screen and 2 weeks for carrier testing. Please contact the laboratory for urgent cases.