immunomodulator (azathioprine, methotrexate or mycophenolate mofetil). Multiple strictures were noted in 25% of patients. Location of strictures included ileal, ileocaecal, sigmoid and caecal.

A total of 32 dilatations were performed in the 20 patients and 8 patients underwent multiple endoscopic balloon dilatations (7 patients underwent 2 dilatations and 1 patient had 6 dilatations). 85% of patients were symptomatic (abdominal pain, vomiting) from the stricture and after endoscopic balloon dilatation in 70.5% the symptoms had improved.

There were 2 procedure related complications 0.06% (1 perforation requiring surgery and 1 perforation was managed conservatively). Mean follow-up since the first stricture dilatation was 2.67 years (0.1-6.11 yrs). During the follow-up of these 20 patients; 4 underwent stricture related surgery and 80% have not undergone any surgical intervention.

Summary and Conclusion Our experience has shown that endoscopic balloon dilatation is a relatively safe procedure for the treatment of luminal strictures. Endoscopic balloon dilatation results in symptomatic relief and delays surgical intervention in Crohn’s patients with luminal strictures.

Case A previously fit and well 13-month-old boy of South Asian descent born to consanguineous parents presented acutely with persistent diarrhoea, vomiting and a perioral rash. He was febrile, developed bloody diarrhoea and clinically deteriorated with significant weight loss (50th centile to 10th centile). He continued to have episodes characterised by cyclical vomiting and feed intolerance, often with associated febrile illness with no microorganisms found from multiple septic screens.

Investigations Investigations showed persistently raised inflammatory markers, anaemia and thrombocytopenia, with hypogammaglobulinaemia. Ultrasound abdomen showed evidence of generalised enteritis initially, but he failed to improve on a course of triple antibiotics. Stool cultures and viral PCR were negative. Faecal calprotectin was raised (>2000 mg/kg). Repeat ultrasound abdomen showed fluid filled, actively peristalsing small and large bowel, with very mild wall thickening and inflammatory mesenteric change. Subsequent upper and lower GI endoscopy was macroscopically normal, and histology revealed chronic active oesophagitis/gastritis with a normal colon.

Ophthalmology assessment due to vision concerns revealed bilateral optic atrophy. MRI brain showed lack of supratentorial, infratentorial and parenchymal bulk with thinning of the corpus callosum and optic nerves.

Progress The rash spread to include the peri-oral, peri-auricular, sacral and genital areas. He was initially treated for acrodermatitis enteropathica due to clinical presentation and borderline low zinc levels, however zinc supplementation did not resolve symptoms. Feed intolerance persisted despite switch to amino acid formula. Parenteral nutrition was commenced after failed enteral (gastric and jejunal) feeding trials.

Whole exome sequencing revealed two missense mutations in the SLC5A6 gene.

Management and Discussion The SLC5A6 gene produces sodium-dependent multivitamin transporters (SMVT) which are expressed in various tissues including the intestine, brain, liver, lung, kidney, cornea, retina and heart. It plays a major role in the uptake of biotin, pantothenate and lipoate in the digestive system and transporting B-group vitamins across the blood brain barrier.

This case was only the fourth to ever be described in literature. The first case described a 15-month-old with failure to thrive, microcephaly, developmental delay, severe immune deficiency and severe gastroesophageal reflux. A subsequent series described two siblings with profound neurodevelopmental, progressive truncal ataxia and refractory cyclical vomiting.

Our patient was managed on vitamin replacement therapy: Biotin (10 mg, intramuscular), Dexpanthenol (250 mg, intramuscular) and α-lipoic acid (300 mg, intravenously) given weekly. With treatment he has shown significant improvement. Cyclical vomiting has settled, his rashes are quiescent, and bloods have normalised. He is now 2 years old, fully enterally fed with his weight on the 70th centile.

This case highlights how defects in multi-vitamin transporters can lead to multi-systemic disease. It also demonstrates the diagnostic role of whole exome sequencing, and with growing genetic databases it will only increase its future potential.

**REFERENCES**