Results The biopsies of the eight sequential patients had been reported by one of four centres within the West Midlands. All, except one, of these local reports were normal. However, subsequent review of all eight biopsies by the national expert identified pathological changes in all. Diagnoses established included two cases of polyglucosan body myopathy, confirmation of NSAID enteropathy and differing forms of inflammation (e.g. lymphocytic plerosis). Establishing these diagnoses enabled accurate prognoses and implementation of subsequent management, including continuation of home parenteral nutrition (HPN, n=6) and consideration for small intestinal transplantation (SIT, n=3). Patients questioned reported additional benefits.

Conclusion Gut dysmotility can be highly symptomatic and debilitating leading to intestinal failure (IF), HPN and SIT. Clinical decisions for consideration for HPN and SIT are complex. Decisions must consider the potential for morbidity and mortality against the potential for improvement in nutritional status, quality of life and survival. A full thickness small bowel biopsy, while invasive, offers opportunity for a definitive diagnosis, and thus a prognosis. Published series report an 81% diagnostic yield for small bowel biopsies in patients with suspected gastrointestinal neuromuscular disorders, when using routine and immunohisto-chemical techniques. However, standard histopathological reporting, which is often based on H&E staining alone, has less potential for achieving a diagnosis. This is shown by our study in which a diagnosis was achieved in only 13%. Thus, our study highlights the importance of expert review and demonstrates the importance of achieving a diagnosis for patient and clinician.

Competing interests None declared.

REFERENCE