Precision medicine in monogenic inflammatory bowel disease: proposed mIBD REPORT standards

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Abstract

Owing to advances in genomics that enable differentiation of molecular aetiologies, patients with monogenic inflammatory bowel disease (mIBD) potentially have access to genotype-guided precision medicine. In this Expert Recommendation, we review the therapeutic research landscape of mIBD, the reported response to therapies, the medication-related risks and systematic bias in reporting. The mIBD field is characterized by the absence of randomized controlled trials and is dominated by retrospective observational data based on case series and case reports. More than 25 off-label therapeutics (including small-molecule inhibitors and biologics) as well as cellular therapies (including haematopoietic stem cell transplantation and gene therapy) have been reported. Heterogeneous reporting of outcomes impedes the generation of robust therapeutic evidence as the basis for clinical decision making in mIBD. We discuss therapeutic goals in mIBD and recommend standardized reporting (mIBD REPORT (monogenic Inflammatory Bowel Disease Report Extended Phenotype and Outcome of Treatments) standards) to stratify patients according to a genetic diagnosis and phenotype, to assess treatment effects and to record safety signals. Implementation of these pragmatic standards should help clinicians to assess the therapy responses of individual patients in clinical practice and improve comparability between observational retrospective studies and controlled prospective trials, supporting future meta-analysis.