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Summary

What can we learn from monogenic IBD

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IBD | Mechanisms & Personalised Medicine

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The speaker reviewed monogenic inflammatory bowel disease, reporting that 1 in 20 pediatric patients in a 1000-patient cohort had a single-gene cause, with potential for curative bone marrow transplant in immune-compartment defects and emerging targeted therapies including SYK inhibitors.

KEY POINTS

- The speaker stated that in a SickKids study sequencing 1000 all-comer pediatric IBD patients, 1 in 20 had a monogenic cause, with higher rates in younger children but occurrence at any age; a literature review reported 10% of adult IBD patients have monogenic disease.
- Monogenic IBD now encompasses over 100 genes; many immune-compartment defects are curable with allogenic bone marrow transplant, while epithelial defects often do not respond to transplant and may require different management.
- The speaker described identifying gain-of-function SYK mutations in patients with severe colitis, B-cell defects, and arthritis; at least 50 patients have been diagnosed worldwide, with bone marrow transplant curative and SYK inhibitors (approved for ITP) showing some efficacy for IBD.
- Autoantibodies to alpha-v-beta-6 integrin were reported in a Japanese study in 90% of ulcerative colitis patients and in a US-Canadian study to predate disease; the speaker suggested these may be causal phenocopies of monogenic disease rather than just markers.
- The speaker recommended considering monogenic panels (now available on exome backbone) for severe adult IBD patients with family history of autoimmune disease, atypical manifestations, or therapy failure, noting that targeted therapies like Anakinra or Abatacept may be appropriate with molecular signatures.

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