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Summary

Approaching the design of low prevalence diseases

Federico Biagi

UEG Week Berlin 2025 | Presentation

Mechanisms & Personalised Medicine | Standards & Guidelines

October 6, 2025

The speaker discusses practical strategies for conducting retrospective multicentre studies in rare small bowel diseases, emphasizing that diagnostic accuracy and careful patient selection are more important than sample size when studying low-prevalence conditions.

KEY POINTS

- The speaker states that all their studies on rare enteropathies including Whipple's disease, atrophic enteropathy, common variable immunodeficiency, and seronegative coeliac disease were retrospective, as prospective studies in rare diseases are very difficult to conduct.
- The speaker cites a prospective Whipple's disease trial from Berlin that enrolled 60 patients over more than ten years, illustrating that sample sizes in rare diseases are inherently small by definition.
- The speaker emphasizes that diagnostic accuracy is paramount and recommends always excluding patients with uncertain diagnoses, because in small samples one misclassified patient can significantly affect results, unlike in larger studies where errors may balance out.
- The speaker categorizes conditions by diagnostic difficulty: Whipple's disease, atrophic enteropathy, common variable immunodeficiency, and coeliac-related malignancies are relatively easy to study due to clear histological patterns, while seronegative and refractory coeliac disease are more difficult, and nonresponsive coeliac disease and autoimmune enteropathy are very difficult due to high risk of false positive diagnoses.
- The speaker recommends selecting expert partner centres that use the same diagnostic criteria, inviting them individually rather than by mass email, keeping them regularly informed, and personally following up with slower collaborators.

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