gutflix
GI learning, personalised

Summary

COMPLEMENT REGULATOR CD55 DEFICIENT PROTEIN-LOSING ENTEROPATHY (CHAPLE DISEASE): SUCCESSFUL TREATMENT WITH POZELIMAB IN TWO SISTERS

Johan E. van Limbergen

UEG Week Vienna 2024 | Poster

Colorectal

October 13, 2024

Two sisters with CHAPLE disease, initially misdiagnosed as Crohn's Disease, achieved sustained remission with pozelimab, the first FDA-approved treatment for this rare complement-driven disorder.

KEY POINTS

- CHAPLE disease is a rare hereditary immune disorder caused by CD55 deficiency leading to complement system overactivation, with fewer than 100 patients diagnosed worldwide.
- Two Moroccan sisters initially diagnosed with Crohn's Disease presented with recurrent edemas, abdominal pain, bloody diarrhea, low serum albumin, and low IgG, refractory to multiple conventional treatments over several years.
- Whole exome sequencing led to CHAPLE diagnosis, confirmed by flow cytometry showing total absence of CD55 antigen on both patients' blood cells.
- Subcutaneous weekly pozelimab treatment resulted in symptom resolution within one month, normalization of inflammatory markers and IgG levels, discontinuation of all immunosuppressive medications within three months, and sustained remission with no related adverse events.
- This case illustrates the diagnostic challenge of CHAPLE disease and the clinical utility of pozelimab, recently approved by the FDA as the first treatment for this condition.

VIEW THIS CONTENT ON GUTFLIX

https://gutflix.eu/search/d/65c5b986-9112-11ef-8acd-0242ac140004

AI-generated summary

This summary was generated by an AI large language model based on the content transcript. It is for informational purposes only and should not be considered a substitute for clinical judgment. Always rely on your professional expertise and the full clinical context when making clinical decisions.