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Summary

Familial Mediterranean fever

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Small Intestine & Nutrition | Surgery | Neurogastroenterology & Motility

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The speaker presented common diagnostic and treatment mistakes in familial Mediterranean fever, emphasizing that FMF diagnosis is primarily clinical and should be considered in patients with recurrent abdominal pain even when other GI diagnoses exist.

KEY POINTS

- The speaker identified as mistake number one the failure to consider FMF as an additional or alternative cause of abdominal pain in patients who already have established GI diagnoses such as IBD or IBS.
- The speaker stated that mistakes two and three involve over-relying on genetic testing to rule out or confirm FMF diagnosis, explaining that the disease is primarily clinical with genetic testing only supportive, and that interpretation should be done only by experienced professionals.
- The speaker reported that FMF is caused by mutations in the MEFV gene encoding Pyrin, with over 400 mutation types existing and autosomal recessive inheritance, though some mutations can produce disease even when heterozygous.
- The speaker described FMF attacks as spontaneously occurring and resolving episodes of aseptic serositis, with systemic amyloidosis affecting predominantly kidneys being the most severe complication potentially leading to end-stage renal failure and death.
- The speaker stated that treatment requires daily lifelong colchicine with a minimum dose of one milligram per day and maximum of three milligrams per day, and identified prescribing subtherapeutic doses and not checking for drug interactions as common mistakes.
- In the Q&A, the speaker described the most distinctive clinical feature as patients being completely incapacitated during attacks with full-blown peritonitis symptoms, then returning to perfectly normal between episodes, and recommended colchicine trial for several months when genetic testing is negative but symptoms fit.

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