Kohlmeier-Degos Disease: A Challenging Case of Systemic Vasculopathy in a 13-Year-Old Female Requiring Novel Therapy

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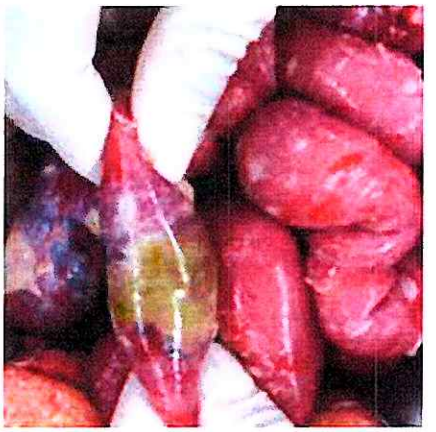
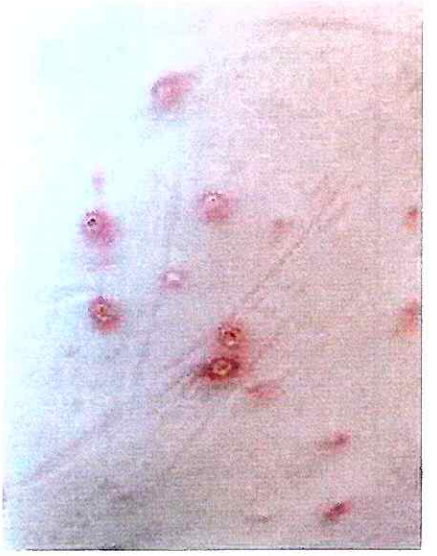


History: A previously healthy 13-year-old female presented with 3 days of abdominal pain, vomiting and rash. Imaging revealed intraperitoneal free fluid and air. Initial laparotomy showed edematous small bowel with patchy ischemia and ileal perforation. Subsequent laparotomy demonstrated significant bowel ischemia and ulceration with diffuse white patches on the small bowel serosa. Exam revealed numerous macules with an atrophic, ivory center bordered by erythema on the trunk and extremities, some with central ulceration and radiating telangiectasias.



Diagnosis and Clinical Course: Clinical findings were consistent with Kohlmeier-Degos (K-D) disease, prompting urgent initiation of eculizumab. Biopsies of skin and small bowel were confirmatory, revealing obliterative fibrosing arteriopathy with microvascular C5b-9 deposits and MXA deposition. Treprostinil was given to mitigate further ischemia. Despite prompt initiation of therapy, she required a small bowel resection. She did well for six months then developed fever, abdominal pain and vomiting. CT imaging revealed a jejunal abscess, consistent with inflammatory enteritis and contained perforation. Exam showed worsening skin lesions. Anifrolumab was added based on initial pathology with MXA deposition and two patient reports with K-D who progressed despite C5-inhibition and benefited from IFN-α blockade. CT showed resolution of the abscess after surgical drainage and IV antibiotics. Skin lesions significantly improved after two doses of anifrolumab.

Conclusion: Systemic K-D disease is a rare, obliterative vasculopathy with characteristic skin lesions and gastrointestinal involvement leading to bowel perforation and death. Timely recognition and initiation of treatment with a C5-inhibitor and treprostinil resulted in prolonged survival for a cohort of patients. A subset of patients, as in this case, will not respond to this combination therapy. Anti-type-1 interferon agents, like anifrolumab, appear to be a promising treatment.KD