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Summary

THE BURDEN OF CEREBROVASCULAR ACCIDENTS IN PATIENTS WITH JUVENILE POLYPOSIS SYNDROME (JPS): WHEN A PREVENTIVE CARDIOLOGICAL WORK-UP CAN BE LIFE-SAVING. A CASE REPORT OF PATENT FORAMEN OVALE (PFO) IN TWO JPS-AFFECTED YOUNG MALE BROTHERS

Alessandro Federico

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Colorectal

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A case report of two brothers with SMAD4 mutation-related juvenile polyposis syndrome presenting with gastrointestinal polyps and patent foramen ovale, one experiencing cerebral stroke, highlights the association between this syndrome and cardiovascular manifestations.

KEY POINTS

- Two brothers aged 24 and 28 presented with multiple gastrointestinal hamartomatous polyps and family history of colorectal polyposis and cancer, with Next Generation Sequencing confirming a SMAD4 gene mutation and diagnosis of juvenile polyposis syndrome.
- The older brother experienced a cerebral stroke caused by patent foramen ovale resulting in permanent neurological impairment, and patent foramen ovale was subsequently diagnosed in the younger brother as well.
- Both brothers underwent percutaneous patent foramen ovale closure to prevent further cardiovascular accidents.
- SMAD4 mutation-related juvenile polyposis syndrome is characterized by extraintestinal manifestations including cardiovascular abnormalities that can lead to acute cerebrovascular events.
- The authors conclude the case highlights the need for preventive cardiological workup in young patients with multiple gastrointestinal tract polyps detected on endoscopy.

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