CLINICAL PHENOTYPE, DIAGNOSTIC ROUTE AND UNIQUE PRESENTATIONS OF PTEN HAMARTOMA TUMOR SYNDROME

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A retrospective study of 53 PTEN Hamartoma Tumor Syndrome carriers in Israel found high rates of malignancies (62%), gastrointestinal polyps (75%), and notable soft tissue tumor prevalence, with mosaicism identified in one case after negative blood testing.

KEY POINTS

- Pathogenic or likely pathogenic PTEN variants were identified in 75% of the cohort (52% missense, 17% nonsense, 25% frameshift mutations), with median age at diagnosis of 38 years
- Malignancies occurred in 62% of individuals, most commonly breast cancer (20%) and thyroid cancer (9%), with three cases of soft tissue tumors including desmoid tumor, osteosarcoma, liposarcoma, and skull-base sarcoma
- Gastrointestinal polyps were present in 75% of patients, five had adenomas, and one had a ganglion cell polyp
- One case of PTEN mosaicism was identified through tissue sequencing (endometrial, salivary, thyroid) after negative blood-based testing
- Five patients (9.4%) received Sirolimus for severe manifestations, with two showing substantial reduction in polyp burden and improvement in gastrointestinal symptoms

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