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An Obese Patient with A Pathogenic *PTEN* Mutation. A Case Report

[Aikaterini Vourdoumpa](#)^{1,2}, [Diamanto Koutaki](#)^{1,2}, [George Paltoglou](#)¹,
[Elena Fryssira](#)³, [Evangelia Charmandari](#)^{1,2} & [Ioanna Bacopoulou](#)¹



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Author affiliations

¹Division of Endocrinology, Metabolism and Diabetes, First Department of Pediatrics, National and Kapodistrian University of Athens Medical School, 'Aghia Sophia' Children's Hospital, Athens, 11527, Greece; ²Division of Endocrinology and Metabolism, Center of Clinical, Experimental Surgery and Translational Research, Biomedical Research Foundation of the Academy of Athens, Athens, 11527, Greece; ³Division of Medical Genetics, National and Kapodistrian University of Athens Medical School, 'Aghia Sophia' Children's Hospital, Athens, 11527, Greece

Introduction: Germline mutations in Phosphatase and tensin homolog (*PTEN*) gene (chromosome 10q23) lead to PTEN Hamartoma Tumor Syndrome (PHTS), which includes Cowden syndrome. *PTEN* encodes a tumor suppressor protein that is a negative regulator of PI3K – Akt signaling pathway, taking part in multiple biological processes, including cellular cycle and regulation of metabolism.

Materials and Methods: We present the case of an 8-year-old female patient followed-up in our Outpatient Obesity Clinic. Clinical examination revealed BMI of 24.8 kg/m² (BMI z-score 2.16), macrocephaly, short neck, nasal base subsidence, lip papillomas and lipoma of the right lumbar area. The past medical history suggested fetal macrosomia, lipoma excision at the age of 4 years and multiple oral papillomas. Clinical genetic evaluation suggested the diagnosis of Cowden syndrome. Genetic testing revealed a *de novo* pathogenic missense mutation of *PTEN* [(NM-0013047175): c.1546-2A>G].

Results: During follow up, thyroid ultrasound showed heterogeneity and two nodules (thyroid function normal, FNA negative for malignancy). Further clinical examination revealed papillomas in the oral cavity and lips and acral keratosis. The BMI initially improved through the implementation of a multidisciplinary personalized lifestyle intervention program, but it gradually increased during puberty (BM: 31.3 kg/m²; BMI z-score 2.95 at 15^{8/12} years). However, despite increased adiposity, our patient did not develop any signs of insulin resistance [3-hour OGTT: Glucose (mg/dL) 0'-89, 60'-119, 120'-114, 150'-120, 180'-109, Insulin (μIU/mL) 0'-8,93, 60'-20,67, 150'-27,81, 180'-19,95 (HbA1c: 5%, HOMA-IR: 1.96)].

Conclusions: PHTS is a rare syndrome that may present early in life and may be associated with dysmorphic features, thyroid nodules and mucocutaneous lesions and obesity but no signs of insulin resistance despite the marked adiposity. Early diagnosis allows the implementation of a multidisciplinary monitoring to detect and manage tumorigenesis on time, as well as other comorbidities. Further research in Cowden syndrome, as a pathophysiologic model of obesity, and increased insulin sensitivity, can potentially provide a deeper understanding of the underlying mechanisms, and contribute to the development of novel therapeutic targets for type 2 diabetes, based on small molecule *PTEN* inhibitors with systematic or tissue specific effects, without oncogenic side effects.

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