Opis choroby *

Definicja

A group rare skin tumor or hamartoma diseases characterized by a germline PTEN mutation and clinical manifestations of hamartomas, overgrowth, and increased risk of neoplasia, notably breast carcinomas, epithelial thyroid carcinomas, endometrial carcinomas, renal cell carcinomas, and colorectal carcinoma. Non-malignant manifestations include macrocephaly, benign thyroid pathology (especially Hashimoto thyroiditis), mucocutaneous hamartomas, colonic polyps, and vascular malformations. Diseases in this group include Cowden syndrome, Bannayan-Riley-Ruvalcaba syndrome, Proteus-like syndrome, Lhermitte-Duclos disease, and Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome.

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
PHTS
PHTS

Kod ORPHA
306498

Kod OMIM
-

Kod ICD10
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Kod ICD11
-

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*Źródło

orphanet