Bannayan-Riley-Ruvalcaba syndrome

Bannayan-Riley-Ruvalcaba syndrome is a rare autosomal dominant disorder caused by a mutation of the tumor suppressor gene PTEN.

It shares the mutation with Cowden syndrome but manifests much earlier. The typical presentation includes macrocephaly and intestinal polyposis, usually hamartomatous polyps. Mild mental retardation is relatively common. There is a temporary delay in motor development, which is corrected later, but myopathies often develop later. Musculoskeletal disorders and skin affections are common. However, the syndrome may represent a risk factor for the development of breast cancer, despite polyposis of the intestine, it is probably not a risk factor for the development of tumors of the gastrointestinal tract.

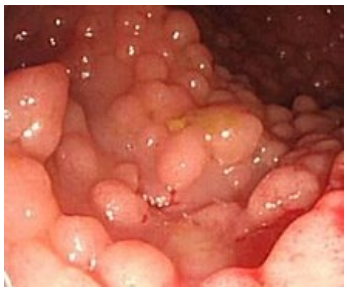
There are also transitional forms of the disease that share the phenotypic features of both Bannayan-Riley-Ruvalcabu syndrome and Cowden syndrome.

Macrocephaly

Links

Related articles

- Cowden syndrome



Intestinal polyposis

Literature

- GEBOES, K. – DE HERTOGH, G.. Non-adenomatous colorectal polyposis syndromes. *Curr Diag Pathol.* 2007, y. 6, p. 479–489, ISSN 1572-0241.
- CALVA, D. – HOWE, J.R.. Hamartomatous polyposis syndromes. *Surg Clin North Am* [online]. 2008, y. 4, p. 779-817, Available from <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2659506/?tool=pubmed>. ISSN 0039-6109.

External links

- Online Mendelian Inheritance in Man. *BANNAYAN-RILEY-RUVALCABA SYNDROME* [online]. [cit. 8/2014]. <https://www.omim.org/entry/153480>.