

Cancer Prone Disease Section

Mini Review

Bannayan-Riley-Ruvalcaba syndrome

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Identity

Alias

Bannayan-Zonana syndrome; Riley-Smith syndrome; Ruvalcaba-Myhre-Smith syndrome; Macrocephaly, pseudopapilledema, multiple hemangiomas; Macrocephaly, multiple lipomas, hemangiomas

Inheritance

autosomal dominant; existence of sporadic cases.

Clinics

Note

Bannayan-Riley-Ruvalcaba syndrome is an overgrowth syndrome/hamartomatous polyposis condition with an increased risk of benign and malignant tumours; other overgrowth syndromes at (known) risk of tumourigenesis are:

- Beckwith-Weideman syndrome,
- Sotos syndrome (cerebral gigantism),
- Hemihyperplasia (hemihypertrophy), and
- Simpson Golabi Behemel syndrome.

Phenotype and clinics

Onset in childhood (in contrast with Cowden disease, although an allelic disorder, see below); more often found in male patients (lower penetrance in female patients).

- Overgrowth at birth (postnatal growth decelerates).
- Macrocephaly.
- Hypotonia and mental deficiency.
- Subcutaneous and visceral lipomas and hemangiomas, and intestinal juvenile polyposis.
- Myopathy of the proximal type in 2/3 of cases.
- Pigmentation spots of the male genitalia.

Neoplastic risk

- Multiple lipomas (75% of cases).
- Hemangiomas (40%).
- Hamartomatous polyps (ileus and colon; 45%).
- Lymphangiomas (10%).

Genes involved and proteins

PTEN

Location

10q23

Protein

Description: 403 amino acids.

Function: Protein tyrosine phosphatase; tumour suppressor gene.

Mutations

Germinal: May be not all Bannayan-Riley-Ruvalcaba syndrome cases are due to PTEN mutations; germ-line mutations have also been described in Cowden disease and in some cases with juvenile polyposis syndrome.

Somatic: PTEN is mutated in a large number of cancer types.

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