



GENETICS AND GENODERMATOSES

## THOSE SPOTS ON HIS PENIS : IT'S BANNAYAN RILEY RUALCABA SYNDROME !!

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**Background:** Bannayan-Riley-Ruvalcaba syndrome (BRRS), an autosomal dominant disorder due to germline PTEN mutation, classically presents with multiple lipomas, hemangiomas, intestinal hamartomatous polyposis, AV malformations, developmental delay, macrocephaly and penile lentiginosis.

**Observation:** A 11 year old boy presented with asymptomatic lesions on lip and tongue Since 2 years, progressive swelling on right thigh since 1 year, and delay in gaining height On cutaneous examination there was presence of:

- Oral papillomatosis (Lip, Tongue, gingiva)
- Acral keratoses
- Palmo-plantar keratotic pits
- Penile lentiginosis (1×1 mm to 3×3 mm)
- Soft subcutaneous swelling in medial aspect of right thigh (15 × 20 cm )

On skeletal Examination:

- Macrocephaly (>2 Standard Deviation)
- Pectus Excavatum
- USG abdomen: Hepatomegaly (14.5cm) with coarse echo texture & Splenomegaly (14cm)
- Colonoscopy: Multiple polyps from rectum to splenic flexure and terminal ileum was seen, histopathology of the polyp was compatible with hyperplastic hamartomatous polyp.
- MRI and CT Angiography showed high flow (Arterio-venous) vascular malformation involving right superficial femoral artery.
- En-block resection of the AV malformation was done followed by full thickness skin grafting by a plastic surgeon and send for histopathological examination showing multiple ectatic thrombosed vessels of varying calibre with arterialisation of vein.
- MRI Brain showed white matter cyst (Parietal Lobe) and increased vascular spaces giving ETAT CRIBLE APPEARANCE.
- With 5 major and 1 minor criteria we made the final diagnoses of PTEN hamartoma syndrome and since penile lentiginosis is characteristic of BRRS, a final diagnoses of same was made.





Key Message: This is a rare disorder and there has been no report of this syndrome from INDIA.

It is usually slow venous malformations which are associated with BRRS, our case had a high flow arterio-venous malformation of superficial femoral artery.

