Assessment of PTEN-associated vascular malformations in a patient with Bannayan–Riley–Ruvalcaba Syndrome

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Disclosure

Speaker name:
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I have the following potential conflicts of interest to report:

☐ Consulting
☐ Employment in industry
☐ Stockholder of a healthcare company
☐ Owner of a healthcare company
☐ Other(s)

☒ I do not have any potential conflict of interest
Clinical findings
Others
The patient, born on 08.01.1989, has been under treatment in our clinic since the year 2002. It is located in the area of the right leg, characterized by a large, mixed, non-spontaneous involuting hemangioma. It consists of a large lymphatic and a smaller capillary component in the sense of a lymphangioma. The finding was first observed in the year 1996 and operated externally. In our clinic, we have performed repeated transarterial embolization treatments in the past.

Furthermore, there is a decompensated knee/ankle joint on the right side with a pronounced Achilles tendon and gastrocnemius muscle shortening as well as a hypoplasia of the right foot (see also reports from the Uniklinik Balgrist).

Further findings include (Thyreoiditis Hashimoto, Macrocephaly of unknown origin, multizystic lesion on the left side paratrigonal) and also orthopedic problems at the contralateral, left leg, which have been reported from the Children's Hospital: St. Osteomyelitis of the left Tibia and a fibrous cortical defect of the Tibial metaphysis. 
Look for

- Macrocephaly
- Penile freckling
- Vascular malformations
- Trichilemmomas
- Over/undergrowth
- Intestinal hamartomatous polyposis

- PTEN-Sequenzing
  (heterozygot missense-Mutation (p.D24Y))
PTEN-Hamartom-Tumor-Syndrom

- Thyroid cancer
- Breast cancer
- Endometrial carcinoma
- Kidney cancer
- Colon-Ca
Surveillance

- Yearly thyroid ultrasound from the time of diagnosis
- Skin check with physical examination
- Annual breast screening and transvaginal ultrasound or endometrial biopsy
- Colonoscopy beginning at age 35 years
- Renal imaging (CT or MRI preferred) age 40 years.
- Family-hx of a particular cancer type > screening 5-10 years prior to the youngest age of diagnosis in the family
Treatment

- Embolization
- Physiotherapy
- Stockings
- Orthopedic shoes

- Sirolimus? Betablocker?
PTEN-Hamartoma-Tumor-Syndrome

- Cowden syndrome
- Bannayan-Riley-Ruvalcaba syndrome
- PTEN-related Proteus syndrome
- Proteus-like syndrome
- High risk for benign and malignant tumors
- Macrocephaly, penile freckling, Vasc.anomalies