Cancer Prone Disease Section

Mini Review

Proteus syndrome

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Identity

**Alias**
Elattoproteus syndrome; Gigantism; Encephalocraniocutaneous lipomatosis; Elephant man syndrome

**Note**
Proteus is a sporadic disorder resulting in a mosaic pattern of tissue overgrowth. The most common manifestations are macrodactyly, asymmetric fat and/or muscle and bone distribution, connective tissue and epidermal nevi, lymphatic and vascular malformations, and hyperostoses.

**Inheritance**
Sporadic

Clinics

**Phenotype and clinics**
General criteria for the diagnosis include mosaic distribution of lesions, progressive course from birth, sporadic occurrence. Supportive findings include connective tissue nevi (characteristically on the soles of the feet and/or on the chest wall), epidermal nevus, disproportionate overgrowth leading to asymmetry. Asymmetric dysregulation of fat in the subcutaneous space lead to unilateral or localized overgrowth and contralateral atrophy of subcutaneous fat. The spine commonly demonstrates scoliosis. Bullous lung disease is often seen and can lead to respiratory insufficiency and pneumonias. Benign tumors include lipomas, lymphangiomas, epidermal nevi, and vascular malformations. There is an increased risk of pulmonary embolism and this is a prominent cause of early death. Cord compression can result from fatty overgrowth into the spinal canal.

**Neoplastic risk**
The risk of malignancy is low. Benign tumors include lipomas, hemangiomas, vascular malformations, lymphangiomas. Neoplasms that have been reported include a mesothelioma, papillary carcinoma of the thyroid, ovarian serous cystadenoma, meningioma, optic nerve tumor and endometrial carcinoma but no systematic risk for particular tumors has been observed.

**Treatment**
Treatment is usually conservative and symptomatic. Surgical repair of severe skeletal distortion is performed although growth is often progressive despite procedures. There is growing concern over the risk of pulmonary embolism although clinical recommendations for prevention have not been established.

**Prognosis**
Longevity is likely affected by Proteus syndrome. A known cause of premature death is pulmonary embolism. Respiratory deaths due to cystic lung disease, massive rib overgrowth and laryngospasm have also been reported. CNS deaths have also occurred from seizures and a cerebellar abscess.

### Cytogenetics

**Note**
No cytogenetic aberrations have been observed in patients with confirmed Proteus syndrome.

**Genes involved and proteins**

**HMGIC**

**Location**
12q15
**Note**
The gene responsible for Proteus syndrome are unknown. This gene, however, is overexpressed in Proteus-associated tumors such as lipomas, salivary gland tumors, leiomyomas and lung hamartomas.

**PTEN**

**Location**
10q23.3

**Note**
The evidence implicating this gene in Proteus syndrome is weak.

**References**


This article should be referenced as such: