Giant Nasal Mass Causing Feeding Difficulty in Tuberous Sclerosis
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What is This?
A 29-year-old woman was referred to us with a huge disfiguring nasal deformity. The patient was intellectually disabled and suffered from epilepsy since birth, thus requiring oral feeding and round-the-clock supervision. Her regular seizures and oligophrenia were thought to be the sequel of perinatal hypoxia. The grape-like nasal mass extended down from her nasal tip and, lately, prevented her mother from feeding the patient properly. Similar skin lesions were also seen on her face (Figure 1). They consisted of groups of papules and nodules, with whitish greasy discharge. Rhinoscopy revealed clear nasal passages. No cervical nodes were palpable. The skin on her back showed areas of thick plaques bulging in a flower-bed fashion. Combined surgical technique of cold-steel dissection, laser vaporization, and monopolar diathermy was used to remove the nasal mass and to establish acceptable cosmetic result. Postoperatively, the patient’s oral functions returned to normal and quality of life improved. Histology revealed giant cell angiofibroma (GCAF) (Figure 2). Immunohistochemical staining (vimentin+, CD34±) confirmed the diagnosis of GCAF. Genetic testing found the presence of abnormal tuberous sclerosis complex 2 gene.

Discussion
Tuberous sclerosis is a multisystemic neurocutaneous tumor syndrome caused by mutation of tumor suppressor genes (TSC1, TSC2)1 causing hamartomas in various organs. Tuberous sclerosis is characterized by the triad of oligophrenia, epilepsy, and sebaceous adenoma. The latter is also called facial angiofibroma2 because of the characteristic histologic appearance of the maculopapular lesions. Shagreen patch in the lumbosacral region is also a typical finding. Other skin disorders, such as hypomelanotic macules, “cafe au lait” spots, and periungual fibromas can be present.3,4 Prognosis is determined by respiratory complications

Figure 1. Tuberous sclerosis causing gross nasal deformity, facial skin lesions, and feeding difficulty preoperatively and postoperatively (upper right corner).

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Keywords
tuberous sclerosis, nose, intellectual disability, feeding difficulty, angiofibroma

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(pulmonary lymphangioleiomyomatosis), intracranial tumors, intractable seizures, and hemorrhages from visceral hamartomas (renal angiomyelolipoma).4,5

Note

The institutional review board of our department approved the manuscript.

Author Contributions

László Lujber, operating surgeon, owner of photographs, writer of the manuscript; András Burián, assistant surgeon, literature reviewer, co-writer of the manuscript.

Disclosures

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References


Figure 2. Giant cell angiofibroma consisting of multinucleated, polymorphic giant fibroblasts (arrows) and various vascular elements (arrowhead).