Giant nasal mass causing feeding difficulty in tuberous sclerosis (case report)

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Tuberous sclerosis (TS) is a rare neuro-cutaneous tumor syndrome with characteristic skin tumors and CNS symptoms.

Disfiguring nasal tumors are also uncommon conditions.

An extreme disfiguring nasal deformity causing feeding difficulty is demonstrated.

Based on the associating symptoms a case of TS unrecognized for 29 years was diagnosed.

In case of characteristic skin lesions and associating CNS symptoms possibility of TS is recommended to be taken into consideration.
Case report

- 29-year-old female patient with intellectual disability, regular seizures
- round-the-clock supervision, feeding
- presumptive cause of intellectual disability and epilepsy: perinatal hypoxia
feeding difficulty

- no intranasal pathology or neck nodes
- to provide per oral feeding & improve QoL
- surgery (general anesthesia): excision (cold instruments & bipolar scissors) + KTP laser
Surgery (video)
Giant cell angiofibroma

HE

vimentin +

vascular element

giant fibroblasts

CD34 (+)
Results
plaques in a flower-bed fashion
Differential diagnosis

malignancy?

rhinophyma?

neuro-cutaneous tumor syndrome?

\[ \text{benign skin tumors} + \ \text{CNS symptoms} \]
Characteristic to tuberous sclerosis!
What is tuberous sclerosis?

- tuberous sclerosis (TS): multisystemic neuro-cutaneous tumor syndrome

- prevalence: 1:6000, AD/sporadical, variable expressivity

- etiology: mutation of tumorsuppressor genes (TSC1 or TSC2) → abnormal proteins (hamartin, tuberin) → disturbed cellular migration and differentiation → hamartomas
classical form: - adenoma sebaceum
- intellectual disability
- regular seizures

characteristic skin lesions + central nervous system symptoms (seizures, intellectual disability, behavioural disorders)

Think of tuberous sclerosis!
Skin symptoms

**Indicators!**

- adenoma sebaceum (facial angiofibroma)
- shagreen patch
- periungual fibromas
- cafe au lait spots
- hypopigmented macules

Images showing typical skin symptoms of the conditions mentioned.
CNS symptoms

- epilepsy

- intellectual disability, autism, ADHD & other behavioural disorders

- in their background: subependymal tumors, nodules, cortical tubers
Visceral manifestations

- **cardial**: rhabdomyoma → heart defect (vitium), conduction problems

- **hepatic**: cysts

- **pulmonary**: lymphangiomatomatosis (respiratory failure)

- **renal**: angiomyolipoma (bleeding)
Diagnosis

- clinical – major and minor symptoms
- genetic test (supplementary)

Coexistence of CNS and skin symptoms

Suspicious for tuberous sclerosis!

A case unrecognized for 29 years…
genetic test (Rotterdam, The Netherlands) → mutation in TSC 2 gene → **tuberculous sclerosis!**

DNA analysis of the relatives: negative → sporadical, postzygotical mutation in the patient
Discussion

Vogt’s triad (adenoma sebaceum, regular seizures, intellectual disability)

2 major symptoms (adenoma sebaceum+shagreen patch)

definitive diagnosis: tuberous sclerosis

- positive genetic test

- giant cell angiofibromas is a rare variant of angiofibromas → cutaneous manifestation is common in TS
Summary

- tumor-like, giant lesions deforming the nose are uncommon
- TS is also rare in ENT field
- the above together are more uncommon
• **ENT relation**: cosmetically and functionally disturbing lesions of the head and neck

• **Prognosis is determined by** the CNS & visceral involvement

**Correct diagnosis is crucial!**
References


Thank you for your attention!

Multidisciplinary approach!