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Clinical Record

Gorlin's syndrome presenting with myolipoma of tongue base

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Abstract

Objective: We report the first case of tongue base myolipoma associated with Gorlin's syndrome.

Method: Case report and review of world literature.

Results: A 39-year-old man with known Gorlin's syndrome presented with progressive dysphagia. Subsequent magnetic resonance imaging scan and biopsy confirmed the rare diagnosis of myolipoma arising from the tongue base. In view of the benign nature of this mass, it was debulked rather than completely excised in order to preserve swallowing function.

Conclusion: To our knowledge, this is the first report of the coexistence of two rare conditions in an atypical fashion. This case appears to represent a new variant in the broad spectrum of features of Gorlin's syndrome.

Key words: Gorlin's Syndrome; Myolipoma; Tongue Neoplasms; Tongue

Introduction

Gorlin's syndrome

Gorlin's syndrome, also known as Gorlin–Goltz syndrome or basal cell naevus syndrome, was first described in 1960. It is an autosomal dominant condition with high penetration[1] and variable phenotype expression.[2] The condition is uncommon, with a low incidence of one in 56 000 persons.[3]

The aetiology of Gorlin's syndrome is thought to relate to chromosomal instability arising in chromosome nine.[4,5] The gene involved is the human homologue of the drosphilia segment gene,[6,7] and has been patched as PTCH1.[8] It is thought that the associated protein has a role in deoxyribonucleic acid maintenance, repair and/or replication,[4] functioning as a tumour suppressor gene in its active state.[9] The condition is usually passed down by familial inheritance, although sporadic cases have been reported.[10,11]

The disorder is characterised by multiple basal cell carcinomas, particularly on the face and at an early age, together with maxillary keratocysts. Other, less frequent clinical characteristics noted in Kimonis and colleagues' series of 82 patients[8] include calcification of the falx cerebri (79 per cent) and tentorium cerebelli (20 per cent), bridging of the sella turcica (68 per cent), abnormal frontal sinus aeration (18 per cent), bifid ribs (26 per cent; most commonly the third, fourth and fifth ribs), calcification of the nuchal ligament (18 per cent), fusion of vertebrae (10 per cent), hemivertebrae (15 per cent), polydactyly (4 per cent),[12] ovarian fibromas (17 per cent of the women), and meningiomas (5 per cent). Other features recorded include palmar or plantar pits,[8] schizophrenia and learning difficulties,[6] retinal hamartoma,[13] and soft tissue tumours such as leiomyosarcoma.[14] Five per cent of patients with the condition develop childhood medulloblastoma. A third of these patients have been shown to have lost the wild type allele of the PTCH gene on chromosome nine, a fact which generated the hypothesis that the PTCH gene was the tumour suppressor gene responsible for the condition.[15]

Malignant features and transformation have also been noted. Khalifa *et al.* reported a case of endometrial adenocarcinoma associated with ovarian fibroma and Gorlin's syndrome.[16] Wolfer *et al.* documented a case of transformation of basal cell carcinoma (BCC) to squamous cell carcinoma in a human immunodeficiency virus (HIV) positive patient with the condition.[17] Yilmaz *et al.* reported the concurrence of basal cell naevus syndrome and adenoid cystic carcinoma in a patient who also had distant metastases in the lungs,[18] a feature seen by other authors in this and other sites.[10,19]

Gorlin's syndrome in otolaryngology

Although not characteristically considered to be associated with otolaryngological pathology, Gorlin's syndrome may affect soft tissue in any part of the body. Hence, ENT cases such as the above salivary gland adenocarcinoma have been reported. Al-Anazy and Zakzouk reported a case of progressive nasal obstruction and hearing loss associated with multiple dental cysts, bifid ribs and calcification of the falx cerebri.[20] Computed tomography scans revealed a large, cystic mass in the left maxillary sinus, protruding into the nasal cavity. The external auditory meati may also be involved,[15,21] and orbital or tarsal BCCs may also extend into and/or obliterate the orbit and ethmoidal sinuses.[8,22] To date, no cases with involvement of the upper aerodigestive tract have been documented.

Case report

A 39-year-old man with a known diagnosis of Gorlin's syndrome presented to the otolaryngology clinic with a

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one-year history of progressive dysphagia, as well as intermittent, left-sided facial pain and otalgia. He had no nasal obstruction, anosmia, rhinorrhoea or otorrhoea, and the nasal, dental and aural examinations were normal. The patient's Gorlin's syndrome had been diagnosed six years previously, when he had had multiple mandibular keratocysts removed. The diagnosis was confirmed had been genetic screening, and an orthopantomogram had shown the facial asymmetry and hemi-mandibular hyperplasia characteristic of the diagnosis. At the time of this previous presentation, the patient had not developed the BCCs usually observed in the condition.

The patient underwent magnetic resonance imaging of the skull base to neck region, which showed a large mass lesion arising from the tongue base (Figure 1).

The patient underwent endoscopy under general anaesthetic, which revealed a large, round lesion arising from the tongue base, with smooth overlying mucosa. The lesion was surgically debulked and sent for histological analysis. In view of its large size, it was not possible to completely excise this lesion without compromising tongue function.

Macroscopically, the specimen comprised firm, rubbery, cream-coloured tissue measuring at least 90 mm in cross section. Microscopically, the tissue consisted of a mixture of adipose and vascular spindle cells (Figure 2). The former were mostly normal, mature adipocytes, but with scattered cells containing multiple globules of fat. The spindle cell component comprised cells with fairly uniform, spindle and ovoid, blunt-ended nuclei and well defined, spindled, eosinophilic cytoplasm. These stained positively with smooth muscle antigen and desmin. They were scattered amongst the adipose tissue but also formed large sheets. Mitoses were not seen, and there was no evidence of malignancy; however, ectatic blood vessels were a prominent component. Mast cells were also seen throughout the lesion. In conclusion, the rare diagnosis of myolipoma was given.

The patient was subsequently followed up at the outpatient clinic. Nine months after surgery, he underwent further debulking of the slow-growing, benign tumour. Complete excision was deemed inappropriate due to the likelihood of impairment of swallowing function.

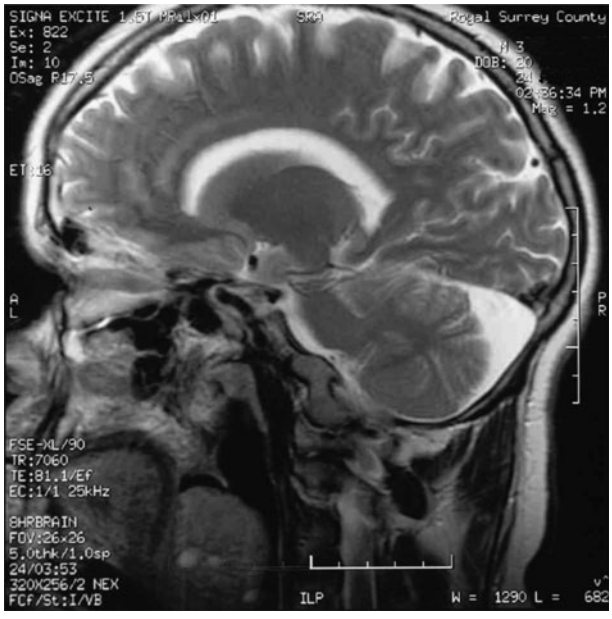

FIG. 1

Sagittal T2-weighted magnetic resonance imaging scan of the patient, showing a large mass lesion in the tongue base.

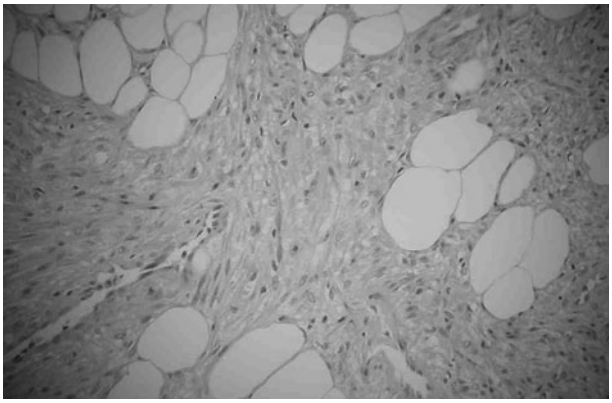

FIG. 2

Photomicrograph showing a mixture of vascular spindle cells scattered amongst normal, mature adipocytes, consistent with myolipoma. No evidence of malignancy was observed (Haematoxylin & Eosin; ×40).

Discussion

Gorlin's syndrome is a rare condition with variable features, of which many variants have been reported. The current report represents the first documented case of an associated tumour of the upper aerodigestive tract. This case is particularly atypical given the lack of BCCs usually so characteristic of the condition.

Myolipoma is also a rare condition, usually affecting the deep soft tissues of the abdomen and/or retroperitoneum. It has not previously been reported in the tongue or pharynx. It is a benign lesion consisting of a variable tissue mass, including smooth muscle fibres containing spindle-shaped eosinophilic cells with cigar-shaped nuclei, and islands of mature adipocytes.[23] No areas of necrosis, mitosis, vascular infiltration or proliferation, or malignant meta- or dysplasia are seen, concurring with the benign nature of these lesions. The present case showed these classical features despite its unusual location. The differential diagnosis for such a mixed tissue tumour includes lipoleiomyoma and fibrolipoleiomyoma.

- **Gorlin's syndrome, also known as Gorlin–Goltz syndrome or basal cell naevus syndrome, is an autosomal dominant condition with high penetration and variable phenotype expression, first described in 1960**
- **The disorder is characterised by multiple basal cell carcinomas, particularly on the face and presenting at an early age, together with maxillary keratocysts**
- **This paper reports the first case of tongue base myolipoma associated with Gorlin's syndrome**
- **The coexistence of these two conditions appears to represent a new variant in the broad spectrum of features of Gorlin's syndrome**

As mentioned, myolipomas are benign and hence carry a good prognosis. In this case, symptomatic resolution was achieved after surgical debulking, and further treatment was therefore considered necessary only for symptomatic control. It should be remembered, however, that malignant transformation of BCCs has been noted in Gorlin's syndrome, albeit in a patient immunocompromised by HIV

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infection. It is of course possible that the present case of myolipoma represents no part of the spectrum of Gorlin's syndrome and is a coincidental finding in a patient with a pre-existing diagnosis. However, as soft tissue tumours have previously been reported in Gorlin's syndrome patients, [14] it seems likely that this case represents yet another variant of this condition, being as it is an oncological syndrome associated with the loss of a tumour suppressor gene.

Conclusion

This case demonstrates the coexistence of two rare conditions in an atypical fashion, and appears to represent a new variant in the broad spectrum of features of Gorlin's syndrome.

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