week of treatment. Several anecdotal reports have illustrated positive results after longer periods of therapeutic levels of steroids: 1 month[9] and 6 months.[10]

Does the finding of positive active temporal arteritis despite steroids suggest insufficient immunosuppression, or that the histological findings do not necessarily correlate with clinical activity? This latter suggestion was made in a paper which took a second biopsy in 20 patients after completion of treatment for temporal arteritis.[11] The group reported that although there was some correlation between the clinical and histological signs of active disease during and after treatment, this was not complete.

The presence of a positive biopsy in this case has confirmed the diagnosis of temporal arteritis. This case at least anecdotally suggests that irrespective of the duration of the steroids temporal artery biopsy may be helpful and should be considered. It is difficult to be confident of the significance of this result in terms of the patient's disease stage. Ultimately, her systemic treatment will be titrated against her symptoms. A positive temporal artery biopsy result when the patient first presented with symptoms suggestive of an arteritis would have confirmed the need for immunosuppression from the outset. This histological confirmation becomes particularly important in patients who develop complications associated with immunosuppression.

References

1. Hunder GG, Bloch DA, Michel BA, Stevens MB, Arend WP, Calabrese LH, *et al*. The American College of Rheumatology 1990 criteria for the classification of giant cell arteritis. Arthritis Rheum 1990;33:1122–8.
2. Hall S, Lie JT, Kurland LT, Persellin S, O'Brien PC, Hunder GG. The therapeutic impact of temporal artery biopsy. Lancet 1983;II:1217–20.
3. Sudlow C. Diagnosing and managing polymyalgia rheumatica and temporal arteritis: sensitivity of temporal artery biopsy varies with biopsy length and sectioning strategy. BMJ 1997;315:549
4. Boyev LR, Miller NR, Green R. Efficacy of unilateral versus bilateral temporal artery biopsies for the diagnosis of giant cell arteritis. Am J Ophthalmol 1999;128:211–5.
5. Ashton-Key M, Gallagher PJ. Surgical pathology of cranial arteritis and polymyalgia rheumatica. Baillieres Clin Rheumatol 1991;5:387–404.
6. Cavallo T. Pathologic approach to the diagnosis of vasculitis. Curr Diagn Pathol 1998;5:70–81.
7. Achkar AA, Lie JT, Hunder GG, O'Fallon W, Gabriel SE. How does previous corticosteroid treatment affect the biopsy findings in giant cell (temporal) arteritis? Ann Intern Med 1994;120:987–92.
8. Allison MC, Gallagher PJ. Temporal artery biopsy and corticosteroid treatment. Ann Rheum Dis 1984;43:416–7.
9. To KW, Enzer YR, Tsiaras WG. Temporal artery biopsy after one month of corticosteroid therapy. Am J Ophthalmol 1994;117:265–7.
10. Guevara RA, Newman NJ, Grossniklaus HE. Positive temporal artery biopsy six months after prednisolone treatment. Arch Ophthalmol 1998;116:1252–3.
11. Fauchald P, Rygvold O, Oystese B. Temporal arteritis and polymyalgia rheumatica: clinical and biopsy findings. Ann Intern Med 1972;77:845–52.

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Sir,

Sphenoid dysplasia and temporal lobe prolapse in neurofibromatosis type 1

The neurofibromatoses are autosomal dominant disorders of the nervous system that primarily affect the development and growth of neural cell tissues resulting in neural, skeletal and dermatological abnormalities. Ocular involvement is manifold and may involve the eyelids, cornea, iris, retina, choroid, optic nerve and the bony orbit. We present a case of an asymptomatic patient with plexiform neuroma in whom ipsilateral sphenoid dysplasia led to herniation of intracranial structures into the orbit.

Case report

A 14-month-old female infant was referred to the eye clinic by a health visitor with a swelling over the left side of the face involving the eyelid. The mother had noticed it at birth and attributed it to the forceps delivery, which had also caused significant bruising of both cheeks. Post-natal history and milestones were normal. There was no significant family history.

Ocular evaulation revealed a visual acuity of 6/7.5 using the Cardiff cards with both eyes open. A non-tender soft tissue swelling with brownish discoloration was noted in the left peri-orbital region. The swelling extended from the lateral aspect of the upper eyelid onto the zygoma (Fig. 1). Anterior segment was normal and corneal reflexes were central. Pupillary responses were

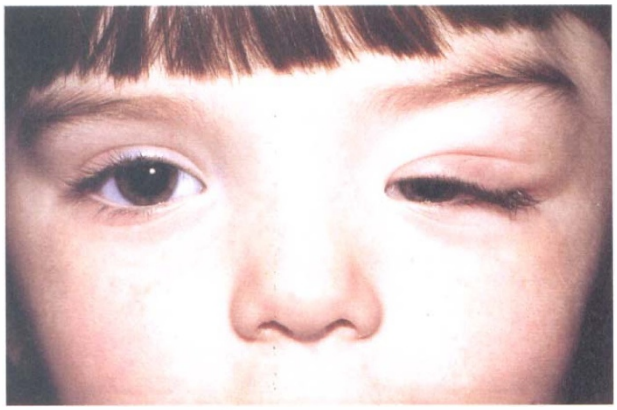

Fig. 1. *Photograph showing the swelling in the left upper lid and the resultant contour of the lid.*

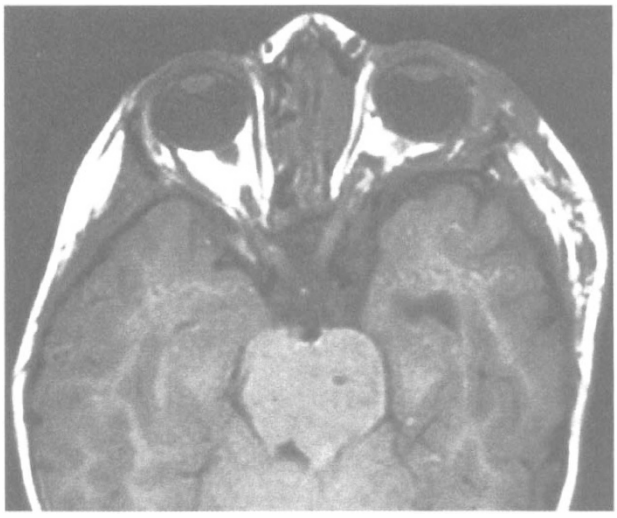

Fig. 2. *MRI of the orbits and brain showing dural ectasia and the prolapsed anterior part of the middle temporal lobe.*

intact. No Lisch nodules were seen. Ocular media were clear with normal fundus examination. A working diagnosis of orbital haemangioma was made with a diagnosis of orbital dermoid and plexiform neurofibroma not ruled out.

Magnetic resonance imaging (MRI) (Fig. 2) of the orbit revealed the swelling to be a plexiform neurofibroma. Deficiency of the greater and lesser wings of the sphenoid were identified (Fig. 3) which allowed dural ectasia and prolapse of the anterior part of the middle temporal lobe into the orbit. Subsequent physical examination revealed two large café-au-lait spots on the right hip and one freckle in the axilla. A diagnosis of von Recklinghausen's disease (neurofibromatosis type 1) was made in the presence of plexiform neurofibroma and sphenoid dysplasia.

In view of the limited cosmetic disability caused by the neurofibroma and keeping in mind the extensive orbito-cranial procedure required to correct the orbital anatomy a decision against surgical intervention was made. Follow-up examination at 3 months showed a decreased visual acuity in the child's left eye. The child

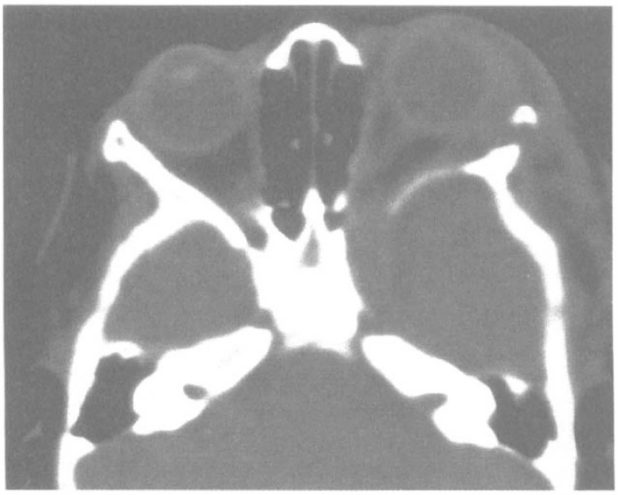

Fig. 3. *Axial CT scan of the orbits showing the hypoplastic greater wing of the sphenoid with dural ectasia and proptosis of the left eye.*

was refracted and started on occlusion therapy for left anisohypermetropic amblyopia. Visual electrophysiology revealed a normal flash electroretinogram and visual evoked response in both eyes. She was treated successfully with occlusion and penalisation. A repeat MRI scan at 6 months revealed no change with the patient remaining asymptomatic and maintaining good and equal vision in both eyes.

Comment

Von Recklinghausen's disease was first described in 1882[1] as a single entity, which was differentiated by the National Institutes of Health[2] in 1987 into neurofibromatosis type 1 (peripheral, classic von Recklinghausen's disease) and neurofibromatosis type 2 (central, acoustic neuromas). Neurofibromatosis type 1 (NF-1) is inherited in an autosomal dominant manner with irregular penetrance and variable expressivity. The responsible gene is located on chromosome 17. The prevalence of this condition is 1 in 3000 and there is no influence of sex, race or ethnic group on its occurrence.[3] The diagnostic criteria for NF-1 laid down by the National Institutes of Health is the presence of two or more of the following: six or more café au lait spots, two or more neurofibromas, plexiform neurofibroma, freckling in the axillary or inguinal regions, optic glioma, two or more Lisch nodules, osseous lesions such as sphenoid dysplasia and a first degree relative with NF-1 by the above criteria.[4]

The diagnosis can, however, pose a challenge, especially as the usefulness of the NIH diagnostic criteria is limited in children younger than 8 years.[5] Cranial features of NF-1 are present only in 3–7% of patients.[6] The presence of a plexiform neuroma should alert one to the possibility of a coexisting sphenoid dysplasia.[6] This can be identified using conventional radiographic study and is particularly useful in cases such as ours where skin and neurological signs are not readily apparent.[7] Sophisticated imaging techniques are used to study the bony orbital morphology. The commonest abnormality is increased inter-temporal distance followed by increased lateral orbital distance and increased medial wall length.

Sphenoid dysplasia is found in less than 4% of cases.[8] The absence of the greater wing of the sphenoid is a distinctive feature of NF-1. Contrary to what was previously believed it is not entirely due to a congenital dysplasia of the sphenoid bone. The only abnormality present at birth can be an enlarged superior orbital fissure and there are progressive changes that eventually lead to the full-blown picture of sphenoid dysplasia.[9] This has also been demonstrated by Harkens and Dolan[10] using serial radiographic, CT and MR images.[10] The absence of the sphenoid wing allows the temporal lobe to prolapse into the orbit.[11] This defect in the membranous portion of the sphenoid can be closed using bone grafts.[11–13] However, the results of this procedure are not entirely sustainable and the bone grafts are known to get resorbed.[11,13] This has led to the use of titanium mesh to cover the defect.[13] Whereas it is important that in cases of

plexiform neuromas an attempt is made to look for sphenoid dysplasia, in the absence of ocular symptoms and signs it is probably reasonable to adopt the wait-and-watch policy.

References

1. Sakai A, Suzuki K. Von Recklinghausen's disease and its pathogenesis. Nippon Rinsho. Jpn J Clin Med 1995;53:2688–90.
2. Ahn MS, Jackler RK, Lustig LR. The early history of neurofibromatosis: evolution of the concept of neurofibromatosis type 2. Arch Otolaryngol Head Neck Surg 1996;122:1240–9.
3. Friedman JM. Epidemiology of neurofibromatosis type 1. Am J Med Genet 1999;89:1–6.
4. Neurofibromatosis conference statement (National Institutes of Health consensus development conference). Arch Neurol 1998;45:575–8.
5. DeBella K, Szudek J, Friedman JM. Use of National Institutes of Health criteria for diagnosis of NF1 in children. Paediatrics 2000;105:608–14.
6. Khairallah M, Messaoud R, Ladjimi A, *et al.* Association of spheno-orbital dysplasia with plexiform neuroma in von Recklinghausen's neurofibromatosis. J Fr Ophtalmol 1999;22:975–8.
7. Abassi-Bakir D, Graiess-Tlili K, Turky A, *et al.* Cranio-facial bone abnormalities in von Recklinghausen neurofibromatosis. Ann Radiol 1995;38:139–44.
8. Kaste SC, Pivnick EK. Bony orbital morphology in neurofibromatosis type 1 (NF1). J Med Genet 1998;35:628–31.
9. Macfarlane R, Levin AV, Weksberg R, *et al.* Absence of the greater sphenoid wing in neurofibromatosis type 1: congenital or acquired. Case report. Neurosurgery 1995;37:129–33.
10. Harkens K, Dolan KD. Correlative imaging of sphenoid dysplasia accompanying neurofibromatosis. Ann Otol Rhinol Laryngol 1990;99:137–41.
11. Havlik RJ, Boaz J. Cranio-orbital-temporal neurofibromatosis: are we treating the whole problem? J Craniofac Surg 1998;9:529–35.
12. Krastinova-Lolov D, Hamza F. The surgical management of cranio-orbital neurofibromatosis. Ann Plast Surg 1996;36:263–9.
13. Snyder BJ, Hanieh A, Trott JA, *et al.* Transcranial correction of orbital neurofibromatosis. Plast Reconstr Surg 1998;102:633–42.

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Sir,

Apocrine sweat gland carcinoma

Apocrine sweat gland carinoma (Moll adenocarcinoma) of the eyelid is extremely rare. The first case report was by Stout and Cooley in 1951.[1] Since then, only seven published cases[2–7] have been consistent with the histopathological criteria for these lid lesions,[2] of which three were illustrated with clinical photographs.[2,5,7] In these reports, the clinical presentation varied between a chalazion-like lid lesion and a blue-red raised nodular mass at the eyelid margin. Apocrine sweat gland carcinoma may spread to regional lymph nodes, and may cause metastatic death.[1,4] The histopathology of this tumour is characterised by an invasive adenocarcinoma with gland-like structures consisting of eosinophilic or opaque glassy cells forming irregularly shaped lumina of varying size.[7]

Case report

An 80-year-old Caucasian man was referred with a history of a rapidly growing painless lump on his right upper eyelid for 2 months. On examination there was an elevated blue-brown soft-elastic tumour, 6 × 4 mm at the medial upper lid margin (Fig. 1). There was notable lash loss from the site of the tumour. The palpebral conjunctiva of the right upper lid showed no abnormalities. Further ophthalmic examination was unremarkable. The regional lymph nodes were not enlarged. His past medical history did not include any malignancy. Full-thickness, wide block excision was performed. Systemic investigation revealed a biopsy-proven well-differentiated carcinoma of the prostate gland (Tlla GI Nx M0). A bone scan was negative for metastasis. The patient was treated for his prostate tumour with a course of hormone therapy. Over a 2 year follow-up there was no recurrent disease of the eyelid or in his regional lymph nodes. His prostate responded well to treatment.

Pathology. Histopathological examination revealed at the eyelid margin a partly cystoid papillary tumour, not in contact with the epidermis, hair shafts or tarsal Meibomian glands. The tumour was composed of cells with a strongly eosinophilic cytoplasm, and moderately pleiomorphic nuclei, containing nucleoli (Fig. 2). The cells showed at several places decapitation secretion, and contained iron-positive, PAS-positive diastase-resistant granules. There was moderate mitotic activity, and

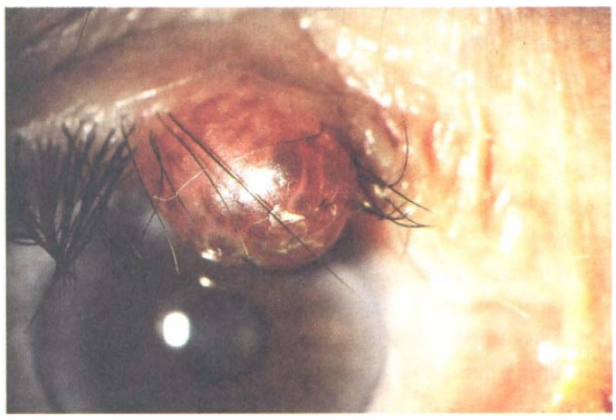

Fig. 1. *Clinical appearance of an apocrine sweat gland carcinoma of the right upper lid of an 80-year-old man. The nodular blue-brown soft elastic tumour measured 6 × 4 mm.*