be designated as another type of chordoma for such lesions.3

Several previous papers have described the differentiation between EP and chordoma. In MRI, addition of Gd-DTPA had no enhancing effects in EP cases, whereas chordomas are commonly enhanced by the addition of Gd-DTPA.1,2 The radiological findings of our case, together with the histological findings of hypocellularity, marked vacuoles and low proliferative activity, might support the diagnosis of EP.

Regardless of the nomenclature, intradural notochordal growth is rare, primarily seen in middle-aged and elderly patients. Only one case has been reported in the paediatric population.1 The number of reported cases is now very limited and the natural history of this condition remains unclear. Long-term follow-up is needed to see if this lesion will develop into typical chordoma showing invasive growth and bone destruction.

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Lipoid proteinosis: a case report

Sir,

Lipoid proteinosis is a systemic disorder of unknown aetiology characterised by deposition of proteaceous material in skin, mucous membranes and internal organs.1–3 Clinically, the disorder is characterised by hoarseness of voice which appears in infancy followed by development of skin lesions.4–6 The disease runs a benign course and is compatible with a normal life span.

We describe an unusual and rare case of lipoid proteinosis along with a brief review of the literature.

A 40-year-old male presented with painless skin lesions on both eyelids developing over a period of 4 months. He had no history of pruritis or other positive symptom related to the lid lesions. The best corrected visual acuity was 6/6, near vision was N/9 both eyes. There was generalised thickening of the skin all over the body with acneiform pick-like scarring on the face (Fig. 1) and extremities. On close inspection of the lid lesions, there were multiple tiny, waxy, skin-colored, flat-topped papules 1–3 mm in size distributed over the entire lid margin on all four lids giving them a characteristic beaded appearance (Fig. 2). Slit lamp biomicroscopy of the anterior ocular segment was normal. The patient also had restricted motility of the tongue. Systemic examination was within normal limits. Investigations including haemoglobin (Hb), total and
differential count, erythrocyte sedimentation rate (ESR), blood sugar and serum creatinine were normal. A definite clinical diagnosis could not be made and it was presumed to be some form of deposition disorder. Biopsy from the eyelid and the scar area on routine H&E staining showed a thinned out epidermis and a thickened dermis filled with amorphous pink hyaline material distributed in the upper dermis around blood vessels, sweat glands and hair follicles (Fig. 3), oriented perpendicular to basement membrane (Fig. 4) that was periodic acid-Schiff (PAS) positive and diastase resistant. A diagnosis of lipoid proteinosis was made with a close differential of amyloidosis which was ruled out with Congo red staining.

A plain radiograph of the skull did not reveal any calcification in the hippocampus area.

Lipoid proteinosis is a rare disorder of autosomal recessive inheritance also called Urbach–Weithe1–3 disease after its first reporters. Usual presentation is in infancy with hoarseness as the presenting symptom, while skin lesions develop within the first 2 years or appear later in life.

Hamada7 has summarised the clinical features of LP in order of frequency as following:
1. Hoarseness of voice.
2. Beaded eyelid papules (monilial blepharosis).
3. Inability to protrude tongue.
4. Ice-pick or acneiform scars.
5. Verrucous nodules with thickening.
6. Blisters and hyperkeratosis.
7. Epilepsy.
8. Alopecia.
9. Gastrointestinal bleeding.

The exact biochemical nature8 of the material has not been elucidated but is thought to be either collagen or carbohydrate-protein complex with unsaturated hydrophobic lipids, hence the name.

Clinically lipoid proteinosis needs to be differentiated from nodular amyloidosis, erythropoietic purpura, lichen myxedematous and colloid milium.

Although no treatment is known to cure the lesions, various drugs have been tried including long courses of steroids, etretinate, D-penicillamine, etc. Removal of the lesions can also be attempted by CO₂ laser or dermabrasion.

Lipoid proteinosis runs a benign course and is compatible with normal life span. The only cause of mortality is respiratory obstruction in infants due to laryngeal lesions.

Our report describes an ophthalmic presentation of a rare and uncommon systemic disorder in a patient of purely Indian origin.

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Molecular testing for paraffin embedded material

Sir,

There has been recent discussion in Pathology over the perceived lack of molecular cytogenetic testing for soft tissue tumours and other paraffin embedded material in Australia.1–3 Our laboratory has been providing this service to many institutions around Australia for several years. The HER-2 fluorescence in situ hybridisation (FISH) laboratory at St Vincent’s Hospital in Sydney was established as the national HER-2 FISH reference laboratory in Australia and is accredited by the National Association of Testing Authorities (NATA) for FISH services. Since 2001 we have successfully analysed paraffin embedded tissue from over 5000 breast cancer cases for HER-2 amplification by FISH. The laboratory is located within the Anatomical Pathology department at St Vincent’s and works almost exclusively with paraffin embedded material. The analysis is performed by two anatomical pathologists with research experience in molecular biology.

Working collaboratively with the FISH laboratory of the Cytogenetics department at St Vincent’s, our services have expanded over the last few years to include paraffin FISH testing for gliomas, soft tissue tumours, lymphomas, and melanomas. We have analysed over 150 gliomas for 1p and 19q deletions and epidermal growth factor receptor (EGFR) amplification. We have also successfully analysed paraffin embedded material from over 100 soft tissue tumours and lymphomas. Our soft tissue probes include dual-colour breakapart probes for rearrangements involving SYT(18q11.2), EWSR1(22q12), CHOP(12q13), and ALK(2p23). Our haematological/lymphoma probes include dual-colour, dual-fusion translocation probes for IgH/MYC, IgH/bcl-2, IgH/CyclinD1 as well as breakapart probes for MYC and ALK genes. FISH can be performed alternatively on tumour touch imprints.

Molecular cytogenetic testing is becoming increasingly relevant. It aids in accurate diagnosis of tumours, imparts prognostic information, and guides therapy in some cases. Currently there is no provision under Medicare to fund these services; however the Department of Veterans Affairs has recently agreed to fund 1p/19q FISH testing in oligodendrogliomas for veterans and their families, upon application. We hope this will lead to more general government financial support for FISH testing as part of the diagnostic workup in select tumours. The Royal College of Pathologists of Australasia (RCPA) has a role to play in liaising with the Medical Services Advisory Committee (MSAC) on this issue.

The St Vincent’s paraffin and haematology FISH laboratories have been providing a reliable and prompt FISH service for several years. We welcome all referrals. A comprehensive list of probes and current pricing may be obtained by emailing the authors or by visiting www.sydpath.stvincents.com.au and clicking the link labelled ‘Information Sheets’.

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