LIPOID PROTEINOSIS: URBACH-WIETHE DISEASE

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Urbach-Wiethe disease (UWD) [1] is a rare, recessively inherited multisystem disorder affecting primarily the skin and mucosae of the "muzzle" region. There is a characteristic deposition of a hyaline material in the arterio-capillary walls and epithelial basement membrane regions, leading to thickening of the skin or mucosae and a predisposition to minor trauma resulting in widespread scarring [2], and which may cause difficulties with tracheal intubation [3].

CASE HISTORY

A 19-yr-old Caucasian male presented for removal of 3rd molar teeth. He was known to have UWD which affected primarily his skin, pharynx and vocal cords. His 21-yr-old sister was affected similarly, but his parents were devoid of the disease. When young children, appreciable laryngeal involvement had limited their voices to a whisper that was high pitched and squeaky in quality. This had necessitated speech therapy, but the patient's voice still remained hoarse and weak.

The patient gave a history of being prone to severe acne which required virtually continuous antibiotic therapy. Examination revealed profuse pitted scarring to the face. Small papules were present along the eyelid margins, which were characteristically thickened. His buccal mucosa was pale and streaky with depressed scars and he tended to scar badly following minor injuries.

Laryngoscopy of the vocal cords had been performed at the age of 4 yr, under general anaesthesia, and the nasal airways were noted to be impassable. At this time biopsy of the vocal cords was performed and the histology reported as UWD. Other patients have undergone similar biopsies with no ill effects and satisfactory healing [4].

Indirect laryngoscopy was performed as part of his preoperative assessment and showed a normal nose, throat and post-nasal space; however, the vocal cords could not be seen. Nasal endoscopy was performed, therefore, and this gave a good view showing the vocal cords to be pale and grossly thickened, the left being both thick and irregular (fig. 1). Both cords were mobile. The patient was otherwise healthy and active; physical examination was unremarkable. He smoked 10 cigarettes a day.

The patient was premedicated with papaveretum 15 mg and hyoscine 0.3 mg. Anaesthesia was induced with thiopentone and the patient was

**SUMMARY**

An otherwise healthy patient with Urbach-Wiethe disease required surgical removal of two 3rd molar teeth. In this multisystem disorder infiltration of the buccal, pharyngeal and laryngeal mucosa may cause difficulties with tracheal intubation and increase the likelihood of trauma. The anaesthetic implications and management are described.

Fig. 1. Photographs taken after induction of anaesthesia using an Olympus LF-1 Fibrescope.
allowed to breathe nitrous oxide and halothane in oxygen via a Magill circuit until the laryngeal reflexes were abolished. Direct laryngoscopy confirmed the preoperative findings and provided an adequate view of the larynx and vocal cords. The vocal cords were sprayed with 4% lignocaine using a Forrester spray. Suxamethonium administered i.v. facilitated atraumatic tracheal intubation with a 7.0-mm cuffed Portex tracheal tube. The pharynx was packed gently. Following return of neuromuscular function, the patient was allowed to breathe spontaneously.

Small areas of gingival tissue (excised with the teeth) appeared to be infiltrated. Bleeding was not noticeably worse than usual for this procedure. Rings of discolouration on the excised teeth were evidence of long term oxytetracycline therapy.

At the end of the procedure, the pack was removed, the pharynx cleared and the trachea extubated with the patient in the lateral position. He made an uneventful recovery and was discharged 2 days later. There was no change to his voice in the early postoperative period or at 3-month follow up. The mouth healed normally. Histology of the gingival tissue taken with the teeth confirmed the diagnosis of UWD.

DISCUSSION

More than 200 cases of lipoid proteinosis have been described in the literature, the majority from northern Sweden and South Africa where the prevalence appears to be greatest. Symptoms depend on the severity of the disease and the structures affected. The skin lesions resemble acne, with small papules particularly along the borders of the eyelids. Hyperkeratosis on regions exposed to mechanical trauma may be present [3].

Hoarseness is the commonest symptom which becomes apparent from an early age [5]. Progressive narrowing of the respiratory tract and development of an immobile "woody" tongue in severe cases may predispose to dysphagia, problems with gustation and mouth opening [3, 6]. Stridor and dyspnoea have not been reported recently, but in 1943 Ramos e Silva reported a case of asphyxia which required prompt tracheotomy [7]. Radiographically, soft tissue films may demonstrate any pronounced laryngeal involvement, although their use in preoperative investigation is of limited usefulness. Biopsies of laryngeal mucosa have demonstrated scanty mucous glands surrounded by deposits of hyaline material [8]. It is postulated that this may account for the diminished lubrication exaggerating the dysphagia and problems of phonation. The reduction in pharyngeal secretions mitigate against the use of antisialagogue premedication. The friable inelastic mucosa dictates that laryngeal instrumentation and tracheal intubation should be as gentle as possible, although protracted haemorrhage has not been reported in these patients [8]. The infiltration of the soft tissues of the tongue and larynx may cause difficulties with direct laryngoscopy and it would be advisable, especially in those with severe disease, to be prepared to use a flexible fibreoptic laryngoscope to visualize the larynx and secure intubation. A reduced gag reflex may occur with this condition [6] and thus care should be taken to avoid aspiration in the postoperative phase by careful attention to pharyngeal toilet and position.

It is said that these patients may have a propensity to develop epilepsy [3, 9] and this may be related to the intracranial calcification [10] well described in UWD. It would seem wise, therefore, to avoid epileptogenic anaesthetic agents.

Reference was made to the fact that lipoid proteinosis is a multisystem disorder. There is said to be an association with, and possible predisposition to, diabetes mellitus [7], although there are no recent corroborative references.

In conclusion, lipoid proteinosis is a rare disorder. As with most disorders of this category, it may be missed unless the physician is aware of the salient features. The anaesthetist should pay particular attention to the abnormal upper airways of these patients. Difficult intubation should be anticipated particularly when there is gross tongue involvement, which is a late manifestation of this disease. Antisialagogue premedication should be avoided. In view of the reported association with epilepsy, anaesthetic agents with epileptogenic properties should not be used.

REFERENCES


