Temporomandibular Joint Degeneration in Alport's Syndrome: Review of Literature and Case Report

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A case of Alport's syndrome and coincidental temporomandibular joint degenerative disease is reported. Alport's syndrome is an unusual genetic disease that ultimately results in renal failure and has a high incidence of sensorineural hearing loss. The patient presented had complaints of facial and joint pain that mimicked what is currently termed temporomandibular disorder, including headache, tinnitus, joint pain, and temporal swelling. The significance of renal osteodystrophy is briefly reviewed as it pertains to this clinical presentation. The clinician must be alert to the many potential causes of degeneration of the temporomandibular joint, one of which may be metabolic in origin.
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Alport's syndrome comprises progressive hereditary nephritis with sensory neural hearing loss. Described in 1927 by Alport, the exact mechanism that destroys the kidney remains unclear, although a defect in the glomerular basement membrane has been suggested.[1–3] The inheritance of the syndrome is unclear as well, but it is most commonly described as being autosomal dominant. There is a male predominance, with end-stage renal failure being reached when the patient is between 20 and 40 years of age.

The first symptom is usually hematuria, often appearing in childhood, coupled with proteinuria. The usual course is one of slow deterioration of renal function. The sensorineural hearing loss may occur at any age, and it primarily affects the high tones and is progressive. A spectrum of ocular dysfunction has been reported, which includes perinuclear anterior lenticonus, perimacular and retinal flecks, and possibly arcus juveniles.[4] Finally, thrombocytopenia has been reported to be somewhat common.[5]

Case Report

A 46-year-old white woman was referred to the Oral and Maxillofacial Surgery Department by her otorhinolaryngologist for evaluation of "TMJ syndrome." The patient's chief complaint was one of facial and jaw pain. In particular, the pain was located over the temporomandibular joints (TMJs) with radiation to the angle of the mandible. The facial pain was accentuated with chewing and eating. The patient also complained of swelling on both sides of her head and associated temporal headaches.

Her past history demonstrated a diagnosis of Alport's syndrome at age 25, with chronic dialysis-dependent renal failure, bilateral

sensorineural hearing loss, and ocular dysfunction. A familial pattern has been demonstrated, as her sister has the same disease to some degree.

Clinical examination demonstrated that the skull was increased in its overall size and that there was mild patchy alopecia. The patient's glasses caused skin indentations in the temples bilaterally. Hearing was decreased to the point where the examiner had to repeat many of the clinical questions and speak in excessively loud tones. The TMJ examination showed that the vertical opening was approximately 50 mm, without deviation. Soft crepitus was present upon palpation over the left TMJ, with mild pain. The right TMJ did not demonstrate significant pain or noise. Lateral and protrusive excursions were essentially normal. Oral examination revealed 1- to 2-mm apertognathia and excessive occlusal wear throughout. The dentition was otherwise in a good state of repair. Palpation of the facial musculature demonstrated bilateral atrophy of the temporalis muscles, but the remaining facial muscle examination was unremarkable.

Radiologic examination included skull films and a panoramic radiograph. Skull films showed a classic "salt and pepper" appearance with increased radiopacity of the calvarium (Fig 1). The panoramic radiograph showed radiopacity in the maxilla and mandible and loss of the lamina dura surrounding the dentition. The mandibular condyles exhibited degenerative joint disease, with the erosion and pitting being worse on the left than the right (Fig 2).

Multiple diagnoses, including Alport's syndrome, renal osteodystrophy, degenerative joint disease, bruxism, and a skeletal malocclusion, were rendered.

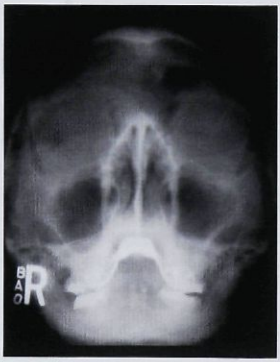

Fig 1 Waters view of skull showing increased radiopacity.

Discussion

In our review of the literature, no case of TMJ involvement has been reported with Alport's syndrome. The incidence of degenerative TMJ is therefore unknown in this syndrome. The obvious underlying pathology of Alport's syndrome is renal failure and its associated systemic changes. Renal osteodystrophy is a generic term used to describe various osseous diseases in uremic patients. In 1973, Sellers et al[6] reported on two cases of condylar degeneration in which the patients were afflicted with severe renal failure and osteodystrophy. This appears to be the first report of condylar degeneration secondary to renal osteodystrophy.

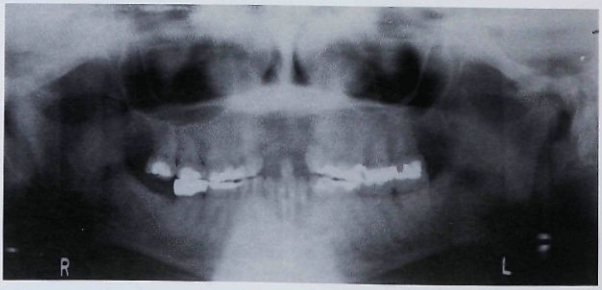

Fig 2 Panoramic radiograph showing increased radiopacity and degeneration of mandibular condyles.

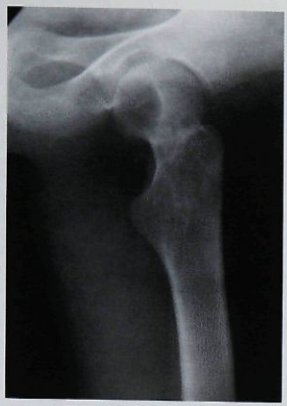

Fig 3 Radiograph of femoral head demonstrating radiolucency consistent with giant cell lesions.

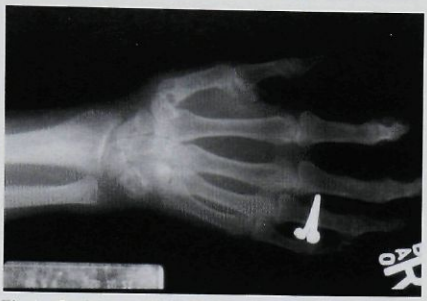

Fig 4 Radiograph of wrist demonstrating radiolucency consistent with giant cell lesions and articular degeneration.

Since patient survival has increased with the development of hemodialysis and transplantation, osteodystrophy has become a greater concern. The pathology of renal osteodystrophy is characterized by increased osteoclastic activity, with resorption and fibrosis caused by changes in calcium and phosphorous metabolism; abnormal vitamin D metabolism; and increased parathyroid activity. Primarily, impaired absorption of calcium and retention of phosphate cause a decreased serum calcium. Secondarily, the hyperactivity of the parathyroid glands causes an increase in parathyroid hormone production, which releases excretion of phosphate, decreases urine calcium excretion, and results in increased release of calcium from the bone.[7,8] More recent information suggests that aluminum toxicity may be an important factor as well.

This complex mechanism results in multiple osseous defects such as resorption, erosion, osteosclerosis, cystic lesions, and giant cell tumors. In this case, osteosclerosis of the skull is apparent, resulting in a classic salt and pepper appearance (Fig 1). The condylar heads of the mandible exhibit erosion and pitting, particularly on the left (Fig 2). Osteosclerosis of the maxilla and mandible, with loss of the lamina dura of dentition, is readily apparent. Finally, cystic lesions and giant cell tumors are apparent in the femoral head and wrist (Figs 3 and 4). Whether the degenerative disease of the condylar head of the TMJ is due to renal osteodystrophy or to chronic microtrauma from bruxism is open to speculation and will require further clinical evaluation of patients with this disease process; however, it appears that the former disease process is most likely causative.

Treatment is directed toward managing the underlying renal disease by obtaining a physiologic balance among calcium, phosphorus, and vitamin D, thus reducing the deleterious effects of renal osteodystrophy. If primary joint pain is paramount, then arthrocentesis or arthroscopic surgery may be beneficial as a lavage and lysis. Splint therapy for bruxism and muscular pain may be therapeutic, but would appear to be of little benefit for metabolic degenerative joint disease. Anterior repositioning splints would be contraindicated, as the meniscal tissues would be in poor condition, malpositioned, or absent in their entirety; splinting in such circumstances would lead to direct condylar loading and result in accentuation of the degenerative process. Physical therapy may be of benefit for muscular pain control. Medications such as nonsteroidal anti-inflammatories would be contraindicated because of their potentially deleterious effects on the kidney. Other medications for pain modulation would have to be individually evaluated for their risk-benefit ratios.

Conclusion

A patient with Alport's syndrome having coincidental degenerative TMJ joint disease was presented.

This patient's symptoms were similar to those of other individuals who complain of pain and dysfunction in the area of the TMJ. The examining practitioner should be aware of the primary underlying defect in Alport's syndrome of end-stage renal disease, and that renal osteodystrophy may produce a spectrum of osseous changes, including frank degeneration of articular surfaces, which will alter conventional treatment protocols.

References

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Resumen

La Degeneración de la Articulación Temporomandibular en el Síndrome de Alport

Se reporta el caso de un paciente afectado por un Síndrome de Alport y por la enfermedad degenerativa de la articulación temporomandibular. El síndrome de Alport es una enfermedad genética inusual que finalmente produce una falla renal y tiene una incidencia alta de sordera neurosensorial. El paciente se quejaba de dolor en la cara y en la articulación, el cual imitaba la los síntomas que se atribuyen corrientemente a los desórdenes temporomandibulares, incluyendo las cefaleas, tinitus, dolor articular y la hinchazón temporal. Se describe brevemente el significado de la osteodistrofia renal en lo que se refiere a esta presentación clínica. El clínico debe estar pendiente de las numerosas causas que pueden producir la degeneración de la articulación temporomandibular, una de las cuales puede tener un origen metabólico.

Zusammenfassung

Degenerative Veränderungen des Kiefergelenkes beim Alport' Syndrom: Literatur-Review und Fallpräsentation

Der Autor beschreibt einen Fall von Alport' Syndrom, einhergehend mit der Degeneration des Kiefergelenkes. Das Alport' Syndrom ist eine seltene erbliche Erkrankung, die letztendlich zum Nierenversagen und zur Innenohrschwerhörigkeit führen kann. Die Patientin klagte über Gesichts- und Kiefergelenksschmerzen, ähnlich wie sie bei einer Myoarthropathie des Kausystems auftreten, ausserdem über Kopfschmerzen und temporale Schwellung. Die Bedeutung einer renalen Osteodystrophie wird erklärt. Der Kliniker muss bei degenerativen Veränderungen des Kiefergelenkes auch an solche metabolische Ursachen denken.