Pearls & Oy-sters: Sequential Bilateral Hearing and Vision Loss With Optic Disc Swelling
Due to Sphenoid Bone Craniofacial Fibrous Dysplasia

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Abstract

Bilateral optic disc swelling is a common finding but rarely associated with multiple cranial neuropathies. In this case, an 18-year-old male presented with subacute sequential hearing loss followed by subacute sequential visual loss. Clinical examination revealed bilateral optic disc swelling. Lumbar puncture revealed a normal opening pressure of 15 cmH₂O. This case discusses a rare but important cause of bilateral optic disc swelling in the context of hearing loss, disequilibrium and a normal CSF opening pressure. An overview of the literature is provided and treatment options are discussed to guide further management of similar cases.

Pearls

Craniofacial fibrous dysplasia may affect the skull base and present with multiple compressive cranial neuropathies.
Papilledema with early visual loss is atypical and should raise concerns for superimposed optic neuropathy.

Any vision, hearing or vestibular changes with craniofacial fibrous dysplasia warrant an urgent evaluation to consider surgical intervention.

Corticosteroids may be considered as an empirical treatment of craniofacial fibrous dysplasia with symptomatic cranial nerve compression if surgical decompression is not possible.

Oy-sters

High level of suspicion is required with review of neuroimaging when the clinical picture does not correlate with initial imaging reports.

Bilateral optic disc swelling due to localized optic nerve compression within the optic canal will have a normal CSF opening pressure.

Bilateral visual and hearing loss in a young adult is often attributed to mitochondrial or toxic etiologies, however, symmetrical compressive cranial pathology must also be excluded.

Extensive investigation is warranted for all cases of suspected idiopathic intracranial hypertension with any additional or atypical features, including a normal CSF opening pressure, rapid visual deterioration, male sex, normal BMI and absence of headache.
Case Report

An eighteen-year-old male presented with a three-month history of gradual left, then right sided hearing loss with tinnitus and disequilibrium. Two months later, he noted progressive painless left, then right visual loss. There was no relevant family or past medical history. Clinical examination revealed grade three optic disc swelling bilaterally which was reported as increased retinal nerve fibre layer thickness due to papilledema on optical coherence tomography. Visual acuity was 0.52 LogMAR (20/66) in the right eye and 1.08 LogMAR (20/240) in the left eye. Romberg’s test revealed a mild body sway but Unterberger’s stepping test was positive, with notable deviation to the right. This helped to localize to a vestibular pathology. Neurological examination was otherwise normal. Audiogram confirmed bilateral predominantly sensorineural hearing loss (Figure 1A). Visual Evoked Potentials revealed bilateral prolongation of the P100 cortical potentials, more so on the left side (Figure 1C). These findings raised the suspicion of a pathology involving second and eight cranial nerves.

Routine screening bloods were all unremarkable. These included ESR, CRP, complete blood count and coagulation screen; liver, renal and thyroid function tests; connective tissue disease screen, ANCA and complement levels; serum electrophoresis; and HIV, syphilis, hepatitis and Lyme serology. The pathergy test to investigate for Neuro-Behçet's disease was negative. Lumbar puncture revealed normal constituents and an opening pressure of 15 cmH₂O.

An MRI brain with contrast was initially reported as symmetrical bilateral optic nerve atrophy with prominence of the perioptic CSF space. The report also commented on an incidental periodontal inflammatory process and prominent bilateral anterior clinoid processes. Genetic investigations were arranged to exclude a mitochondrial disorder given the subacute sequential bilateral hearing and visual impairment. Testing for Leber hereditary optic neuropathy, Kearns-Sayre syndrome,
MELAS (Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like episodes) and MERRF (myoclonic epilepsy with ragged red fibers) all returned negative.

An inflammatory optic papillitis was also considered. The patient received empirical pulsed intravenous, followed by oral steroids and the visual acuity improved (Snellen: Left 20/80 from 20/200, right 20/40 from 20/120). Further review of the MRI imaging with the clinical history revealed bilateral stenosis of the optic canals leading to CSF distension of the optic nerve sheaths and clinical picture of papilloedema and visual impairment. This was secondary to hyperostosis of the anterior clinoid process (Figure 2A). There was also stenosis of internal auditory canals due to bony hyperostosis of the petrous temporal bones with compression of the seventh and eighth nerves. A CT scan and subsequently a dedicated bone SPECT scan later confirmed polyostotic fibrous dysplasia involving the skull, skull base and mandible, and also excluded any malignant process. Pituitary hormone function tests were all unremarkable. These included prolactin, growth hormone, IGF-1, cortisol, ACTH, LH and FSH.

The patient underwent an urgent endoscopic bilateral optic nerve decompression. Visual acuity initially deteriorated significantly postoperatively despite highly satisfactory post-operative appearances on imaging. This was possibly due to postoperative swelling of the optic nerves resulting in a localized compartment syndrome in the remaining portion of the optic canal. A prompt second endoscopic procedure was undertaken to remove the remaining portion of sphenoid bone extending distally to the optic chiasm. Abdominal adipose tissue was then inserted at the site of decompression to prevent CSF leakage. Visual acuity three days following the second procedure was 1.26 LogMAR (20/360) at one meter in the right eye and just hand motion, close to the face, in the left eye. Visual acuity one month (38 days) following the second procedure was 1.06 LogMAR (20/230) in the right eye and 1.56 LogMAR (20/720) in the left eye. A repeat audiogram after 32 days was reported as normal (Figure 1B). This suggested improvement in hearing with a combination of
decompressive surgery and high dose corticosteroids which were gradually tapered over the next three months.

Discussion

We present a rare case of craniofacial fibrous dysplasia causing bilateral hearing and visual loss with optic disc swelling which initially improved slightly with corticosteroids. We believe this to be the first case described in the literature of craniofacial fibrous dysplasia causing symptomatic bilateral compression of both the optic and vestibulocochlear cranial nerves. Fibrous dysplasia is a non-neoplastic congenital process resulting in the replacement of bone with an abnormal fibrous tissue. The condition can be monostotic or polyostotic, affecting either single or multiple bones respectively, with a clear predilection for the long bones, ribs and craniofacial bones. Most lesions are usually incidental, monostotic and asymptomatic, requiring only clinical observation and patient education.

At a molecular level, fibrous dysplasia is associated with an activating mutation in the GNAS1 gene which is located at 20q13.2-13.3 and encodes for the alpha subunit of G protein (guanine nucleotide-binding protein).\(^1\) G protein helps stimulate adenylate cyclase, which in turn regulates the production of various hormones and also the signaling pathway for osteogenesis. Interestingly, fibrous dysplasia is not inherited as the gene mutation invariably occurs after conception. Lesional tissue biopsy has the highest sensitivity (80%) in comparison to serum lymphocytes for PCR-based genetic diagnostic methods (20-30%).\(^2\)
Craniofacial dysplasia of the skull base has a wide variety of presentations including atypical facial pain, headache, sinus issues, proptosis, diplopia, strabismus, visual changes, hearing loss and facial numbness. Bisphosphonates have been trialed for pain reduction and to possibly slow the rate of growth but further studies are required. Malignant transformation is very rare and transformation to osteosarcoma and other sarcomas occurs in less than one percent of cases.

A single-center case series of ten patients with craniofacial fibrous dysplasia identified reduced visual acuity in two individuals, one with a coexisting contralateral vestibular syndrome, and objective hearing loss in another individual. A further review of 66 cases of fibrous dysplasia affecting the temporal bone identified the two most common presenting complaints to be headache (59%) and hearing loss (29%). In another case series, thirty patients with fibrous dysplasia affecting the clivus presented with either headache or cranial nerve deficits; these manifested with a variety of symptoms including tinnitus, vertigo, disequilibrium and diplopia.

The clivus (Latin for slope) lies near the center of the brainstem and may be affected by a variety of cystic, neoplastic, meningeal, infective, traumatic and dysplastic pathologies. Surgical approaches have previously relied on microsurgical transcranial techniques from the anterior, posterior and lateral directions however endoscopic transnasal transclival approaches are becoming increasingly favored. The management of symptomatic craniofacial fibrous dysplasia is not established but decompressive surgery is often considered. A comprehensive meta-analysis of 241 fibrous dysplasia patients with 86 clinically impaired optic nerves confirmed a post-surgical improvement in visual function in around two thirds, but a statistically significant post-operative visual deterioration for asymptomatic nerves. This risk of deterioration was attributed possibly to an increased sensitivity of the already damaged optic nerve to surgical insult. Long term follow up of asymptomatic nerves encased with fibrous dysplasia has suggested a low risk of optic neuropathy, especially in the
absence of growth hormone excess which is a statistically significant risk factor and may suggest a pathophysiological relevance.\textsuperscript{10}

Our case initially demonstrated a measurable improvement in visual acuity with corticosteroids. The only reported similar improvement involved a 22-year-old female with unilateral visual loss and optic neuropathy that initially improved with orbital decompression. A further recurrence of vision loss in the absence of any radiological changes fifteen months later then responded completely to a tapering does of oral prednisolone. She was reported to remain well for nine months following the discontinuation of steroids.\textsuperscript{11} The exact mechanism for this sustained improvement remains unknown. Corticosteroids may exert their effect by either slowing the proliferation of fibrous tissue, or reducing any localized optic nerve edema. Review of our case and others in the literature highlights the risk of post-surgical deterioration. This could be mitigated by advocating unilateral decompression of the most symptomatic nerve in the first instance. The role of empirical corticosteroids remains unclear but we would recommend considering their use as an emergency bridging strategy peri-operatively, or for lesions that are unamenable to surgery.

References


Figure 1. Audiograms and Visual Evoked Potentials
(A) Audiogram demonstrating sensorineural hearing loss at presentation. (B) Satisfactory audiogram one month (32 days) following the second procedure while still receiving corticosteroids. (C) Pre-operative Visual Evoked Potentials showing bilateral prolongation of the P100 cortical potentials, more so on the left side. Average P100 latencies were 165 ms on the left and 135 ms on the right.
Figure 2. Radiological findings and postoperative changes.

A. Axial T1-weighted image showing hyperostosis of the anterior clinoid processes with bilateral optic canal stenosis. B. Axial T2-weighted image showing bilateral CSF distension of the optic nerve sheaths with cupping of the optic nerve heads. The CSF distension only extends posteriorly up to the optic canals. C. Axial CT Head with bony windows showing bilateral optic canal stenosis. D. Coronal CT reconstruction showing bony hyperostosis with ‘ground glass’ changes in the central skull base, typical of fibrous dysplasia. E. Axial CT scan showing bilateral bony hyperostosis of the petrous temporal bones with stenosis of the internal auditory canals. F. Heavily T2-weighted MRI scan of the Internal auditory meati showing bilateral compression of the seventh and eighth cranial nerves. G. Post-operative Axial T1-weighted and T2-weighted (H) images show bilateral decompression of the optic canals bilaterally with persistent CSF distension of the optic nerve sheaths.
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