OCT Documented Optic Nerve Neuropathy in Pycnodysostosis - Case Report and Review of Literature

Neelam Pawar1*, Meenakshi Ravindran1, S Padmavathy2, Devendra Maheshwari3 and R Ramakrishanan3

1Paediatric Ophthalmology and Strabismus Department, Aravind Eye Hospital, Tirunelveli, India
2Neuroophthalmology Department, Aravind Eye Hospital, Tirunelveli, India
3Glaucoma Department, Aravind Eye Hospital, Tirunelveli, India

*Corresponding Author: Neelam Pawar, Pediatric and Squint Services, Paediatric Ophthalmology and Strabismus Department, Aravind Eye Hospital, Tirunelveli, India.

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Abstract

We report the case of a 10-year-old girl who presented to us for ophthalmic evaluation referred by pediatrician. On fundus examination, disk pallor was noted in both eyes more in left eye. She did not complain of headache, vomiting or blurred vision. Computed tomography (CT) imaging showed diffuse thickening of skull vault and base with hyperostosis along with narrowing of optic canal and skull base foramina. Spectralis-OCT imaging revealed optic neuropathy in both eyes. This case report reviews the clinical and radiographic characteristics of pycnodysostosis and OCT documented optic neuropathy.

Keywords: Pycnodysostosis; Optic Neuropathy; OCT; Mandibular Angle

Introduction

Pycnodysostosis is a rare genetic disease characterized by osteosclerosis and bone fragility first described in 1962 by Maroteaux and Lamy. It is characterized by short stature, increased density of the bones (osteosclerosis/osteopetrosis) and brittle bones [1-5]. The head is usually large compared to body, the nose beaked, the mandibular angle obtuse, and both maxilla and mandible hypoplastic. Other features may include small tip of the fingers with absent or small nails, an abnormal clavicle, frontal, occipital bossing and dental abnormalities [6-8].

We report a case with the typical clinical and radiological characteristics of the Pycnodysostosis associated with an optic atrophy, an association rarely reported. To our best knowledge OCT documented optic atrophy in Pycnodysostosis has not reported in literature.

Case Report

A 10-year-old school going girl was brought to our hospital by parents for ophthalmic evaluation referred by pediatrician. She had history of pain in hips during walking and complain bowing of legs while sitting. The child was born by first degree consanguineous marriage at term by Caesarean section. Birth history was unremarkable. She had history of seizures. She had a birth weight of 2.25 kg and no developmental delay. Family history was negative for ocular abnormalities. The child was of short stature 111 cm, head circumference was 55 cm, weight was 20 kg, BMI 16.23.

On examination, her best corrected visual acuity was 20/20 in each eye (20/60 1.5DC@ 105 in right eye and 1.00DC@ 60 in left eye). Slit-lamp examination was normal for both eyes. Dilated fundus examination revealed optic disk pallor with normal vessels in both eyes.

Intraocular pressure by noncontact tonometry was 14 mm Hg in the right eye and 15 mm Hg in the left eye. Color vision (36 plate Ishihara) and visual field testing (Bjerrums Screen) were normal in each eye.

All biochemical results, including blood glucose, serum electrolyte (calcium), renal function, and liver function test were within normal limits except phosphorus which was elevated 5.6 mg/dl. USG abdomen showed hepatosplenomegaly. Parathyroid was normal 26.7 pg/ml. TSH was elevated 5.38 and she was on thyronorm 12.5 mg. She had received blood transfusion before presenting to us. She had history of multiple dental procedures.

The girl was short in height, head was slightly large, the nose beaked, the mandibular angle obtuse, and both maxilla and mandible hypoplastic (Figure 1a-1c). She had small fingers and toes.
Examination of the mouth revealed caries of the teeth, impacted and malposed teeth, persistent deciduous teeth and missing teeth. X ray showed obtuse mandible angle and crowded teeth (Figure 2a). Computed tomography (CT) imaging showed diffuse thickening of skull vault and base with hyperostosis along with narrowing of optic canal and skull base foramina (Figure 2b). Dilated fundus examination revealed optic disk pallor with normal vessels in both eyes (Figure 3a and 3b). The clinical and radiological features exhibited by this patient led to a diagnosis of pycnodysostosis.

Peripapillary retinal nerve fiber layer (RNFL) thickness measured by spectral domain optical coherence tomography (Spectralis, Heidelberg) confirmed fundus findings of optic neuropathy. Her average RNFL thickness was 51 mm in the right eye and 39 mm in the left eye.

**Discussion**

Pycnodysostosis is a rare genetic lysosomal disease accounting for 1 - 1.6 per million cases. It is characterised by short stature, brittle bones, underdevelopment of the tips of the fingers, an abnormal collarbone, distinctive facial features including a large head, underdeveloped facial bones, dental abnormalities [1-8].
**Figure 2:** (a) Lateral skull radiographs showing narrow mandible, obtuse mandibular angle, crowded teeth  
(b) CT scan showing hyperostosis.
Pycnodysostosis is rare autosomal recessive condition caused by mutations in the gene that codes the enzyme cathepsin K (CTSK) on chromosome 1q21. Though the diagnosis of pycnodysostosis is based on physical features and X-ray findings but molecular genetic testing is often confirmatory. Most of cases require oral and maxillofacial intervention frequently [6-10].

The differential diagnosis is established with osteopetrosis, cleidocranial dysplasia and idiopathic acro-osteolysis [5]. A CTSK gene mutation analysis is the gold standard confirmatory test but it was not done in this case because of its high cost. Exceptionally, hepatosplenomegaly and hematologic alterations have been observed in some cases of pycnodysostosis similar to our patient [5].

We documented a spectral domain documented OCT, which to the best of our knowledge, has so far not been reported in cases of pycnodysostosis. Particular attention was paid to monitor patient periodically for possible surgical intervention if narrowed cranial foramina caused symptoms like headache and visual loss.

OCT has proved a valuable imaging modality to monitor patients with fibrous dysplasia for development of optic neuropathy during periods of conservative watchful waiting [10].

Craniosynostosis, frequent fractures, respiratory-sleep problems, and dental problems may cause significant morbidity in patients of pycnodysostosis [3,8]. The clinical heterogeneity of this disease and its rarity makes it difficult to provide patients an accurate prognosis, appropriate care, counselling and follow-up [8]. Prevention through proper counselling and periodic follow up is essential in eliminating factors that predispose patients to risk of visual loss and ensuring better quality of life.

Visual acuity of 20/20, with preserved visual fields, may be seen in a patient with optic atrophy. Her average RNFL thickness in both eyes was less than normal Indian paediatric age [11].

This case report highlights that OCT can be used in clinical practice in cases with diagnostic uncertainty to clarify the presence of ONH neuropathy in rare cases where there no obvious sign and symptoms of increased intracranial pressure and optic nerve compression exists.

**Conclusion**

We suggest an active screening of all patients diagnosed with Pycnodysostosis for ophthalmological evaluation as a new clinical feature of this disorder.

Case management should involve a multidisciplinary team representing ophthalmology, neurology, and interventional radiology.

**Disclosures**

The submitted material has not been previously published and is not under consideration for publication elsewhere.

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Authors have read the final manuscript within their respective areas of expertise and participated sufficiently in the study to take responsibility for it and accept its conclusions.

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**Bibliography**


