Case Report

Painful proptosis and compressive optic neuropathy in an 11-year-old girl with tuberculosis

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Keywords

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Abstract

An 11-year-old girl presented with sudden onset of unilateral proptosis and periocular pain. Initial examination revealed proptosis, restricted vertical ocular motility with vertical binocular diplopia, decreased visual acuity and visual field impairment in the left eye. Systemic work-up revealed extrapulmonary tuberculosis with an inflammatory lesion at the left orbital apex causing a compressive optic neuropathy (CON). Three months after treatment with anti-tuberculosis (TB) therapy the proptosis regressed, the ocular motility normalized, and the central visual acuity recovered. The optic atrophy persisted with partial loss of the visual field. This is a very rare presentation of tuberculosis in an 11-year-old child.

Case Report

We report the case of an 11-year-old girl presenting at the University Hospitals Leuven with one week history of pain in the left eye and a progressively increasing left proptosis in the last month (fig. 1). There were no other general symptoms (no fever, weight loss or nocturnal sweating). The patient had immigrated from Somalia 3 years before.

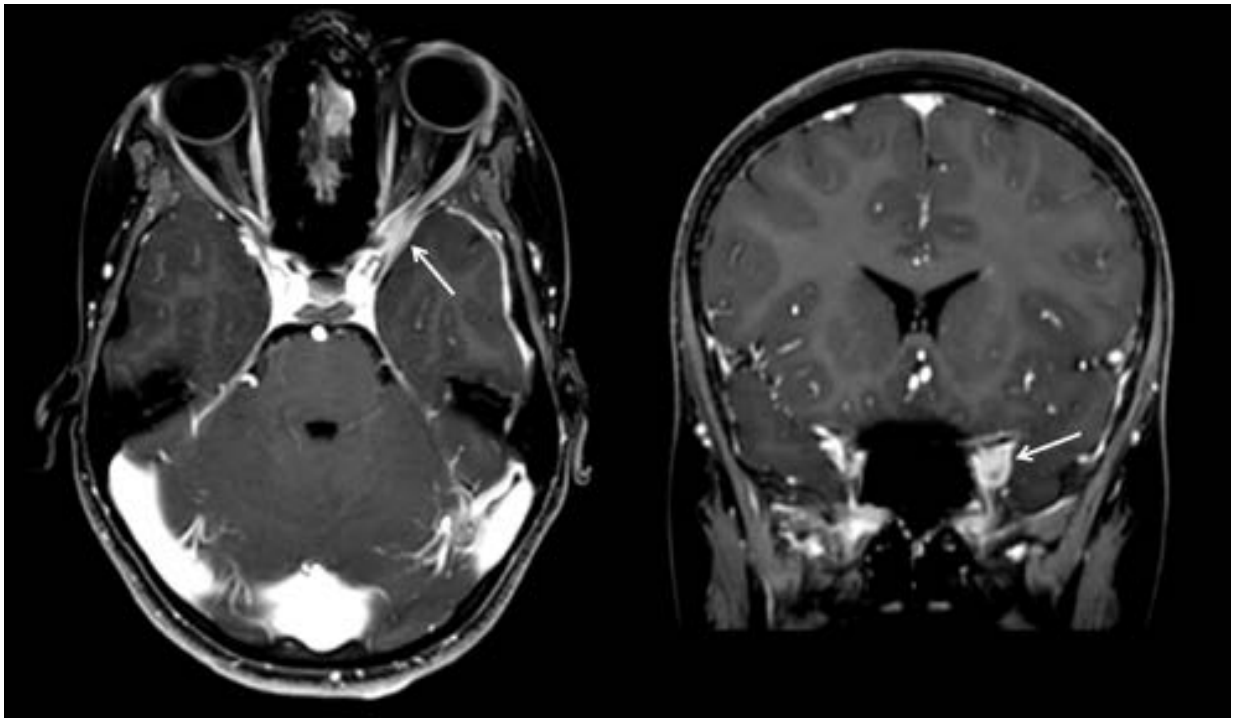
At a first examination, elevation and depression of the left eye were restricted which induced binocular vertical diplopia. Central visual acuity was normal but visual field defects with globally reduced sensitivity were seen. Minimally impaired colour vision, a left relative afferent pupillary defect (RAPD), associated with moderate optic disc pallor, were suggestive for an optic neuropathy. Somatic examination showed no signs of meningism, no fever, no skin lesions. Pulmonary, abdominal and cardiac auscultations were normal and there were no abnormal systemic neurological findings. The only non-ocular pathological clinical finding was the presence of a palpable right abdominal soft tissue mass.

The cranial MRI showed an unsharply delineated intra-orbital lesion with a diameter of +/- 1 cm at the apex of the left orbit (fig. 2), causing a mass-effect and inducing proptosis and a compressive optic neuropathy (CON) without infiltration into the optic nerve, as well as two nodular lesions at the left lamina cribrosa with expansion into the ethmoid sinus. Whole body positron emission tomography and computed tomography (PET-CT)) showed a pulmonary lesion in the left apex, two nodular lesions in the right kidney, a distal cortical left femoral lesion, mediastinal and abdominal lymphadenopathies were also visualized.

Purified protein derivative (PPD) skin test and interferon gamma release assay (IGRA) were both positive. A first biopsy of a mediastinal lymph node by mediastinoscopy was not conclusive but showed no arguments for a lymphoma (which was the first diagnosis to exclude in the differential). The bone marrow biopsy was normal. Before considering more invasive surgical biopsy of the abdominal lymph nodes, a transtracheal echo-guided subcarinal lymph node biopsy was performed. Polymerase chain reaction (PCR) and culture of

Figure 1 : *Left proptosis*

Figure 2 : *T1-weighted images with contrast (Gadolinium), note the contrast enhancing lesion at the left orbital apex (arrow)*



these samples were positive for Mycobacterium tuberculosis, sensitive to all first-line anti-tuberculosis drugs. Lumbar puncture was negative. There was no known family history of tuberculosis but the patient and her family were referred to the Flemish Association for Respiratory Medicine and Tuberculosis Management (VRGT) for contact tracing.

Treatment for extrapulmonary tuberculosis was prescribed, with rifampicin – isoniazid – ethambutol – pyrazinamide for 2 months, followed by bi-therapy (rifampicin - isoniazid) for 7 months. Three months after the start of the tuberculostatic therapy there was a clear clinical and radiologic regression of the orbital and extra-orbital lesions. The proptosis relative to the right eye decreased from 3 to 1mm with improvement in ocular motility and resolution of the vertical diplopia. Visual acuity fluctuated during the first month but was ultimately preserved. The visual field improved slightly (possibly due to a learning artefact) but did not fully recover and the left relative afferent pupillary defect persisted, as expected due to the optic nerve atrophy (fig. 3).

Discussion

The most frequent etiology of unilateral proptosis is thyroid eye disease (TED) in the adult, and infectious cellulitis in children (1). Any pathologic change in the orbit needs to be investigated swiftly and thoroughly as due to the proximity of the globe and the optic nerve, significant damage and visual loss can occur even with "benign" orbital diseases (1). Especially when a child presents with a sudden onset of proptosis urgent work-up is indicated as this sign is mostly associated with severe pathological changes and urgent therapy is often required. Rhabdomyosarcoma is the most common primary orbital malignant tumor in children and will present with a rapidly increasing proptosis (1-4). Other malignant and benign causes of paediatric proptosis, in decreasing frequency, are optic nerve gliomas, metastases of neuroblastoma, orbital neurofibromas, vascular malformations such as capillary hemangiomas, metastatic Ewing's sarcomas, dermoid cysts, chloromas of acute myeloid leukemia, Burkitt lymphomas, Langerhans cell histiocytosis, retinoblastomas (only very severe cases expanding through the sclera beyond the eyeball) and arteriovenous/lymphatic malformations (2,3,8). Proptosis with spontaneous periorbital ecchymosis (raccoon eyes) is especially suspect for malignancies (2). Inflammatory diseases such as thyroid eye disease can also cause proptosis in children (2-4).

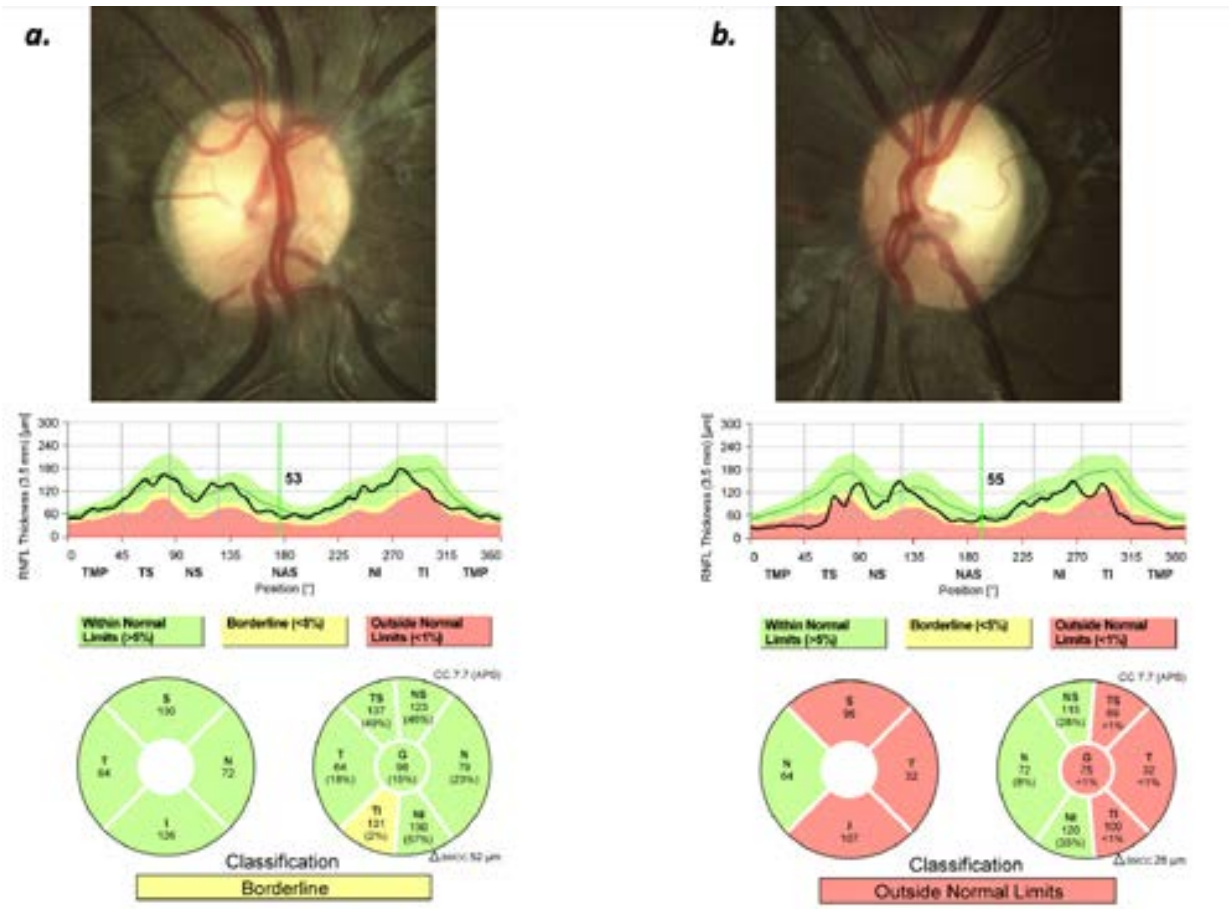
The presence of extra-orbital multiple disseminated lesions throughout the body associated with the country of origin of our patient guided us towards a possible diagnosis of tuberculosis, which was later confirmed.

The incidence of tuberculosis in Belgium is decreasing since the 1990's but the decrease has slowed down more than in our neighboring countries over the last 25 years. In 2019 the general yearly incidence was 8,5/100.000 inhabitants (4,3/100.000 in Belgian citizens versus 38,9/100.000 for the non-Belgian population). For children between 0 and 14 years of age the incidence is 1,3/100.000 in Belgian compared to 10/100.000 in non-Belgian children (5). These discrepant numbers show the impact of immigration on the risk of developing tuberculosis.

When treating patients with tuberculosis it is important to keep in mind that toxic optic neuropathy can be caused by ethambutol or isoniazid toxicity. The most common ophthalmic manifestations of these toxic optic neuropathies are dyschromatopsia and scotomas (6). When symptoms occur, exposure to these drugs needs to be interrupted as soon as possible because damage to the optic nerve can be permanent. In our case no drug toxicity occurred.

Intra-orbital tuberculous lesions, although very rare in Europe, have been described before. Oakhill et al. published a case report in 1982 about orbital tuberculosis in an 11-year-old girl in the UK. She made a complete recovery when treated with anti-TB medication (7). She presented with proptosis but there is no description of visual loss or optic neuropathy. A few other cases were reported in Egypt and India. Hughes et al. published a case series of 7 adults (aged 24-44) who presented with orbital apex syndrome or optic neuropathy attributed to tuberculosis in the UK. Only 2 cases presented with proptosis and 1 with diplopia. All patients showed good visual recovery after treatment with high dose corticosteroids and anti-TB medication (8). Tenawade et al. described an orbital apex syndrome caused by tuberculosis in a 16-year-old adolescent girl in the UK. There was no proptosis but restricted motility and partial ptosis of the upper lid. MRI findings showed an infiltrative lesion around the optic nerve with crowding

Figure 3 : *Colour photograph and Heidelberg Optical Coherence Tomography (OCT) retinal nerve fiber layer analysis a. healthy right optic disc – b. atrophic left optic disc*



at the apex leading to a compressive optic neuropathy. The logMAR visual acuity dropped from 1.5 at presentation to perception of light 9 days later. The visual acuity did not recover and marked optic atrophy was noted 6 months after presentation (9).-

Although the central visual acuity was preserved in our patient, the optic nerve became atrophic due to longstanding compression with a permanent lasting relative afferent pupillary defect and visual field defects. Thanks to prompt diagnosis and treatment further damage could be prevented. No corticosteroids were started in our case as the visual acuity was conserved at the time of the exact diagnosis. Steroids could have masked a lymphoma if started immediately.

To the best of our knowledge, the combination of both proptosis and compressive optic neuropathy due to tuberculosis has not yet been described in a child.

The aim of this article is to report this unusual paediatric presentation of tuberculosis and to emphasize the importance of swift work-up in proptosis to prevent possible permanent visual loss.

Informed consent

Informed consent for publication of this case was given by the patient and her guardian.

Disclosure

None of the authors has a conflict of interest to disclose.

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