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RESEARCH ARTICLE

WHEN ALL TESTS ARE NORMAL: STEROID RESPONSIVENESS AS A KEY DIAGNOSTIC ARGUMENT FOR MULTIPLE SCLEROSIS IN PEDIATRIC OPTIC NEURITIS

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Abstract

Background: Pediatric optic neuritis (ON) represents a diagnostic challenge, particularly when extensive investigations fail to identify a clear etiology. While imaging and laboratory findings are central to diagnosis, clinical reasoning and therapeutic response may play a decisive role.

Case presentation: We report a 14-year-old girl presenting with acute unilateral visual loss of the right eye associated with headache and pain on eye movement. Clinical examination revealed anterior optic neuropa thy with stage II optic disc edema.A comprehensive ophthalmologic, neurological, infectious, and immunological workup was performed. Brain and orbital MRI showed inflammatory involvement of the right optic nerve without evidence of disseminated lesions. Cerebrospinal fluid analysis and extensive autoimmune and infectious testing were unremarkable.

Management and outcome: High dose intravenous methylprednisolon eresulted in rapid and marked visual recovery, with resolution of optic disc edema.

Conclusion: This case highlights that, in the absence of definitive paraclinical evidence, the diagnosis of demyelinating optic neuritis may be suggested but cannot be confirmed, and should rely on careful clinical interpretation and follow-up. Steroid responsiveness may support an inflammatory demyelinating process but is not specific to multiple sclerosis.

Introduction:-

Optic neuritis (ON) is an inflammatory disorder of the optic nerve frequently associated with demyelinating diseases, particularly multiple sclerosis (MS). In adults, diagnosis is often supported by magnetic resonance imaging (MRI) and cerebrospinal fluid (CSF) abnormalities demonstrating dissemination in space and time.However, pediatric ON differs in presentation and often lacks definitive paraclinical markers at initial evaluation. In such cases, the diagnostic process becomes more complex, requiring a careful integration of clinical, radiological, biological, and therapeutic data.Importantly, a rapid and significant response to corticosteroid therapy is recognized as a characteristic feature of inflammatory demyelinating optic neuritis and may contribute to the diagnostic orientation toward MS.However, this response is not specific to multiple sclerosis and may also be observed in other

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inflammatory or idiopathic optic neuropathies.We report a case of pediatric optic neuritis in which all investigations were inconclusive, and the diagnosis relied primarily on clinical findings and a striking response to corticosteroids.The objective of this report is to highlight the role and limitations of steroid responsiveness in the diagnostic approach of pediatric optic neuritis.

Ethical approval and consent:-
Written informed consent was obtained from the patient's legal guardians for publication of this case report. The study adhered to the principles of the Declaration of Helsinki.

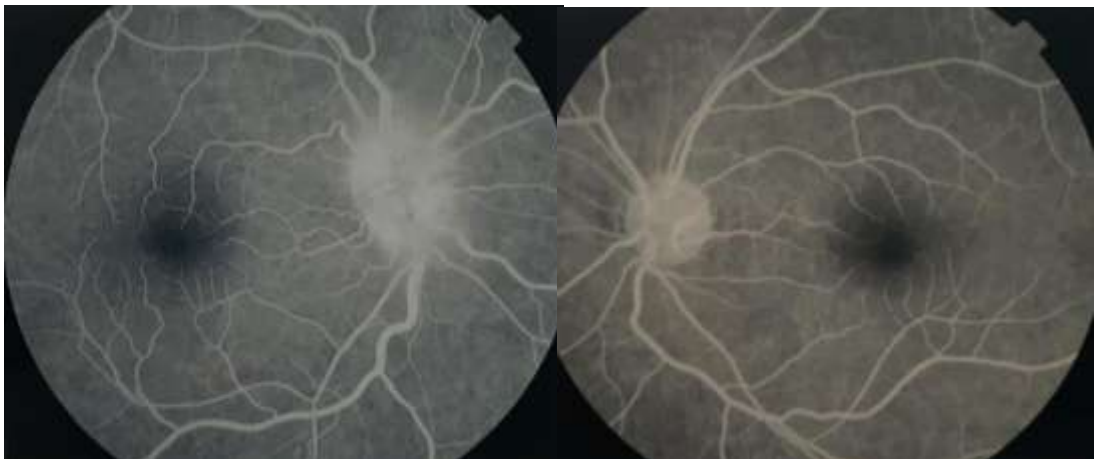
Case Report:-
A 14-year-old girl presented with acute visual loss in the right eye evolving over five days. The condition was associated with headache and pain on ocular movement.

Medical history:-
- Iron deficiency anemia for 18 months (poor compliance with treatment)
- Menstrual cycle disorders (hypermenorrhea and irregular cycles)
- Refractive amblyopia in the left eye
- Recent upper respiratory tract infection 10 days prior

Clinical Examination:-
- **Visual acuity:**
  - Right eye: counting fingers at 2 meters (non-improvable)
  - Lefteye: 5/10
- **Pupillary reflex:**
  - Relative afferent pupillary defect in the right eye
- **Intraocular pressure:** normal bilaterally
- **Anterior segment:** normal
- **Fundus examination (right eye):**
  - Stage II optic disc edema
  - Blurredmargins
  - Papillaryhyperemia
  - Venous dilation and tortuosity
  - Absence of hemorrhages or exudates
- **Lefteye:** normal

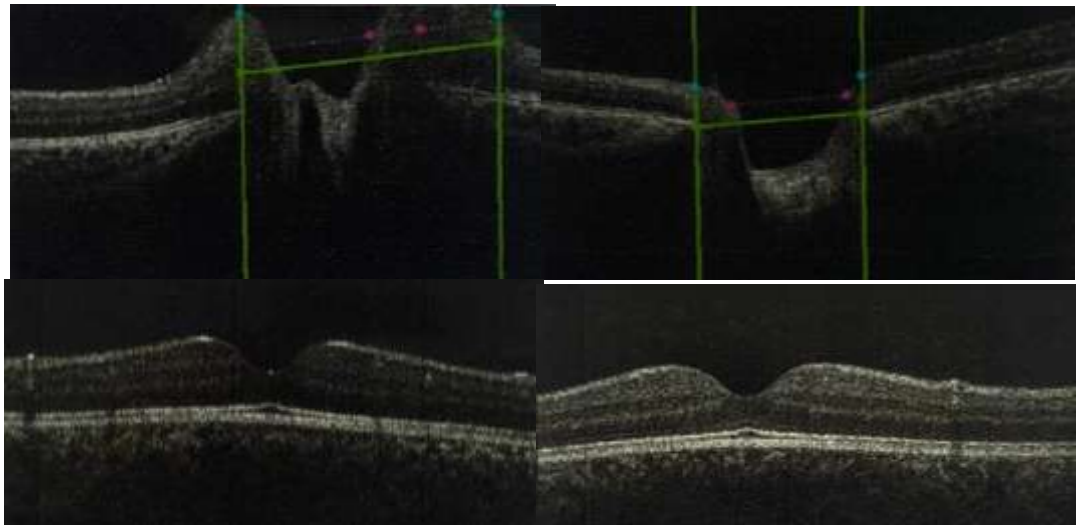
Ophthalmologic Investigations

Fluorescein Angiography:-
- Papillaryleakage
- Peripapillary diffusion
- Venous dilation
  → confirminginflammatoryopticneuropathy



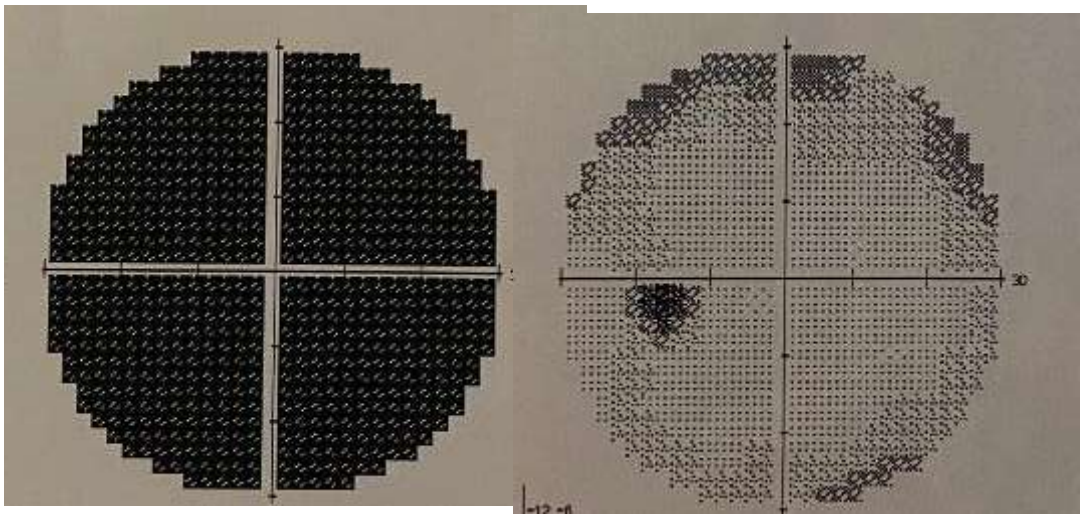
Optical CoherenceTomography (OCT):-

- Increasedoptic disc thickness
- Reduction of physiologicalcupping
- Peripapillary RNFL thickening
- Macular OCT: normal



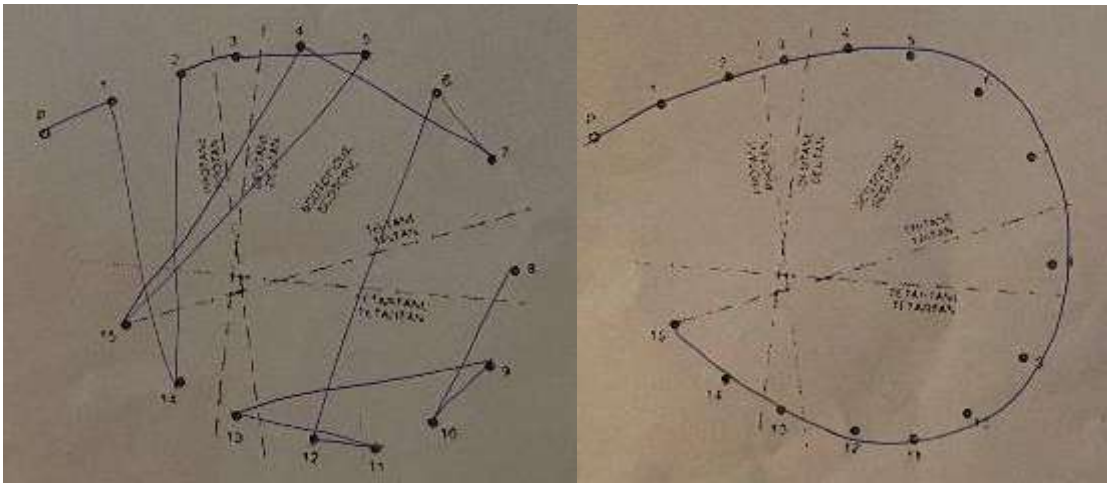
Visual Field:-

- Right eye: non-interpretable due to low visual acuity
- Left eye: learning test, arciform superior defect (to be confirmed)



Color Vision (Lanthony desaturated test):-

- Blue-yellow (tritan) deficit
- Red-green (deutan) deficit
  → consistent with optic nerve dysfunction

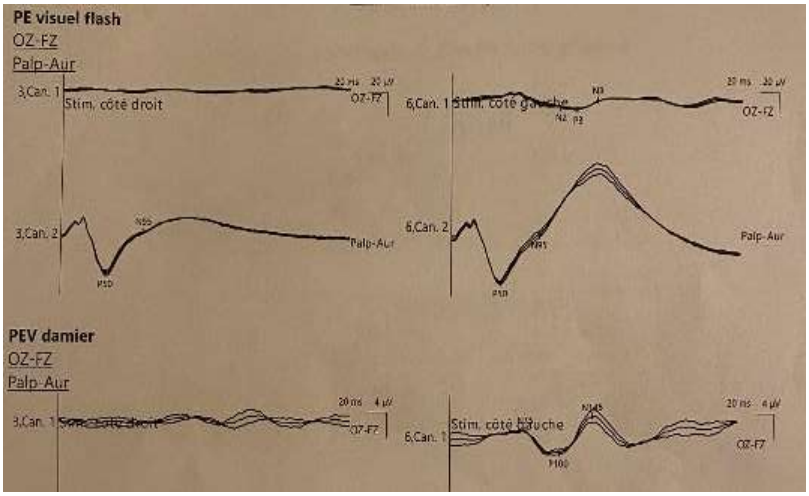


Visual Evoked Potentials (VEP):-

- Right eye: abolition of P100 response
- Left eye: delayed latency and reduced amplitude

→ suggesting:

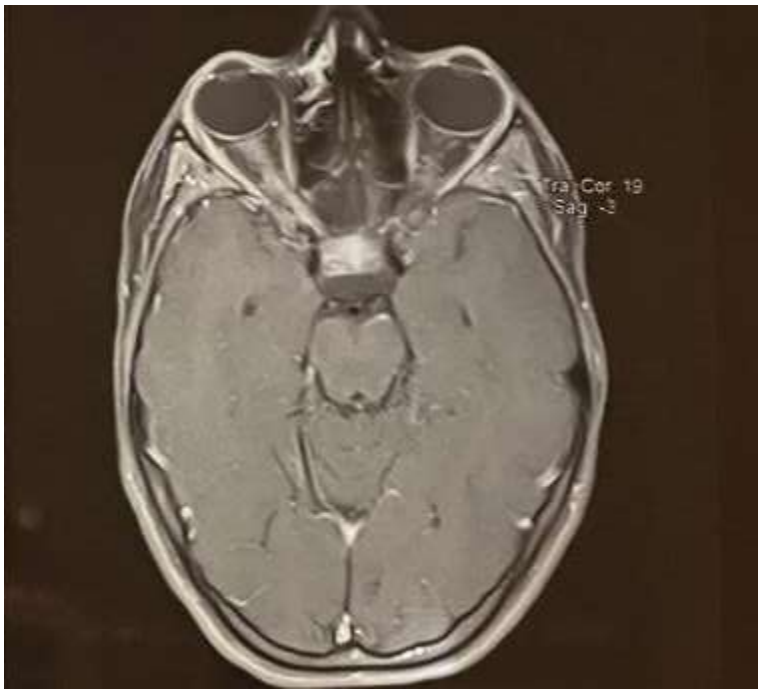
- Severe pre-chiasmatic involvement (right eye)
- Subclinicalinvolvement (lefteye)
- Possible demyelinatingprocess



Neuro imaging:-

MRI (brain and orbits):-

- T2 and FLAIR hyperintensity of the right optic nerve
- Gadolinium enhancement
- Involvement of intraorbital, retrobulbar, intracanalar, and prechiasmatic segments
- No demyelinatinglesionselsewhere
- No compressive lesion



Laboratory and SystemicWorkup:-
Cerebrospinal Fluid:-
- Normal cytology
- Normal protein and glucose
- Negative culture

Biological Workup:-
- Mild anemia (Hb 10.7 g/dL)
- Elevated ESR (52 mm/h)
- Normal CRP
- Lowferritin

Infectious Workup:-
- HIV, HSV, CMV, EBV: negative or immunized
- Syphilis, Lyme, toxoplasmosis: negative
- Tuberculosis (Quantiferon): negative

AutoimmuneWorkup:-
- ANA, anti-dsDNA, ANCA: negative
- Anti-MOG: negative
- Anti-AQP4: negative

Differential Diagnosis:-
- Demyelinating optic neuritis (MS, NMOSD)
- Infectiousopticneuropathy
- Autoimmunesystemicdisease
- Compressive opticneuropathy
- Hereditaryopticneuropathy
- Idiopathic inflammatory optic neuritis

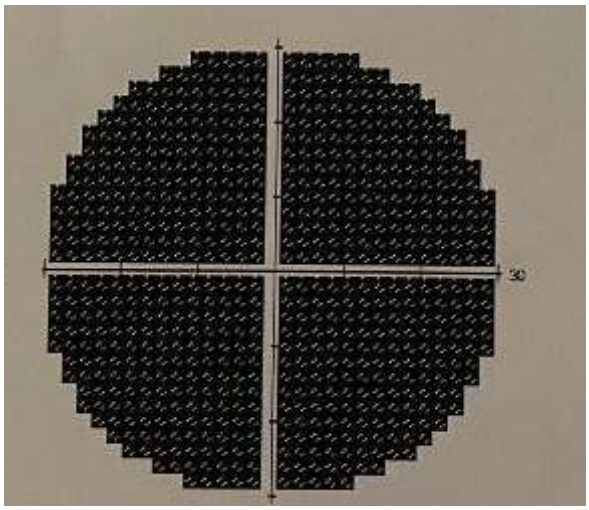
The exclusion of alternative diagnoses was based on the absence of infectious markers, negative autoimmune antibodies, normal CSF findings, and absence of compressive or infiltrative lesions on MRI.

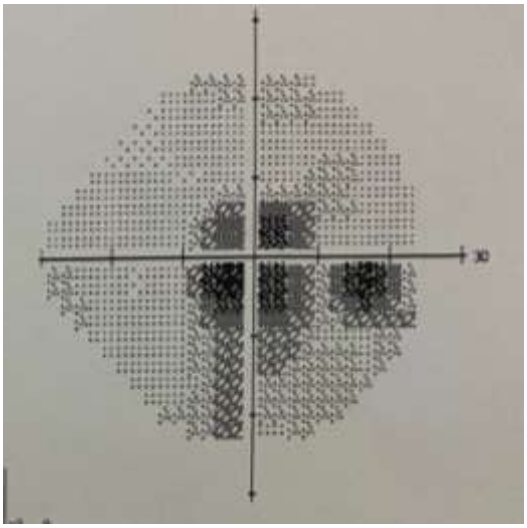
Management:-
- Oral antibiotics for associatedsinusitis
- Intravenous methylprednisolone (1 g/day for 3 days)
- Oral corticosteroidrelay

Outcome:-
Clinical evolution:-
- Visual acuity improved from CF → 9/10
- Pupillary reflex normalized
- Resolution of optic disc edema

Visual field:-

- Improvementwithresidual central scotoma





Discussion:-

Pediatric optic neuritis is often diagnostically challenging due to the absence of typical radiological and biological markers at onset.

In this case:

- MRI did not demonstrate dissemination in space
- CSF was normal
- Autoimmune and infectious workups were negative

However, several clinical arguments strongly suggested a possible demyelinating etiology:

- Acute unilateralvisualloss
- Pain on eyemovement
- Optic disc edema
- Relative afferentpupillarydefect
- Bilateralelectrophysiologicalinvolvement

Importantly, although the rapid response to corticosteroids strongly supports an inflammatory mechanism, it cannot be considered specific for multiple sclerosis. Similar responses may be observed in idiopathic optic neuritis or other inflammatory conditions.Therefore, steroid responsiveness should be interpreted as a supportive but non-specific diagnostic element.Another major limitation in this case is the absence of long-term follow-up. Without clinical or radiological progression, the diagnosis of multiple sclerosis remains hypothetical and requires longitudinal monitoring.This highlights the importance of follow-up MRI and neurological evaluation to assess dissemination in time, which is essential for confirming MS diagnosis. Furthermore, differential diagnoses such as idiopathic optic neuritis and post-infectious inflammatory neuropathy must be carefully considered and cannot be definitively excluded in a single acute episode. This report is limited by its single-case design, lack of long-term follow-up, and absence of specific biomarkers confirming multiple sclerosis. Therefore, conclusions must remain cautious.

Conclusion:-

This case illustrates the limitations of paraclinical investigations in pediatric optic neuritis.
Although clinical features and steroid responsiveness suggest an inflammatory demyelinating process, they are not sufficient to establish a definitive diagnosis of multiple sclerosis.The diagnosis should remain provisional and requires long-term follow-up with repeat clinical and radiological evaluation.This case emphasizes the importance of integrating clinical reasoning with cautious interpretation of therapeutic response.

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