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CASE REPORT

Bilateral Disc Edema in Familial Mediterranean Fever

KUMAR S

ABSTRACT

I report a case suffering from Familial Mediterranean fever (FMF) who presented with complaints of blurred vision. This report illustrate that possible diagnosis of pseudotumor cerebri should be considered as a cause of poor vision in FMF.

Introduction

Familial Mediterranean fever (FMF, MIM 249100) or Recurrent polyserositis [1],[2] is an autosomal recessive multi-system disease, with incomplete penetration [3], manifested by self limited episodes of fever accompanied by painful attacks affecting the abdomen, chest or joints, and sometimes a skin rash [4]. It is most prevalent in people of Armenian, Sephardic Jewish, Levantine Arabic and Turkish ancestry. It was first recognized as a distinct nosological entity only in 1947 [5]. It is caused by mutations in gene MEFV, on the short arm of chromosome 16. Ocular involvement is rare in FMF. Episcleritis [6], optic neuritis, retinal colloid bodies [7], ophthalmoparesis and prolonged latencies of the visually evoked potentials [8] have been reported in FMF.

Case Report

A 23-year-old, single, Arab female, of Palestinian ancestry presented in the eye department of Mohammad Dossary hospital with the complaints of not seeing well with her current glasses for last few weeks. Her best corrected visual acuity was 6/9 and 6/12 in right and left eye respectively.

She read all the Ishihara plates with each eye. Slit lamp examination of both anterior segment was unremarkable. Both pupils were reacting to light without any afferent pupillary defect.

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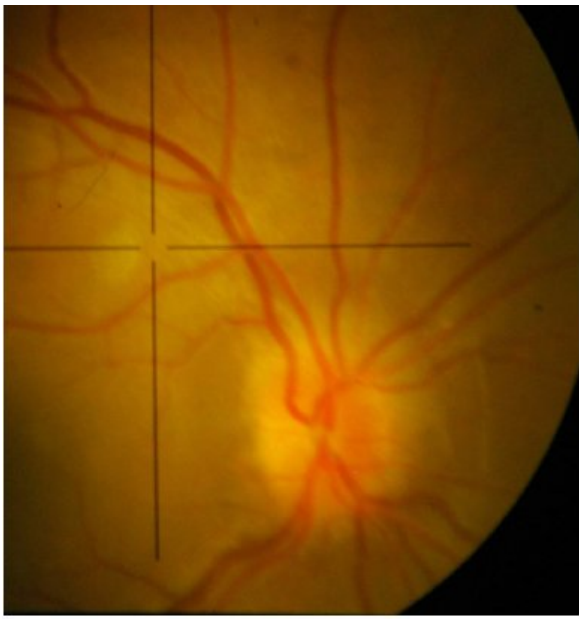
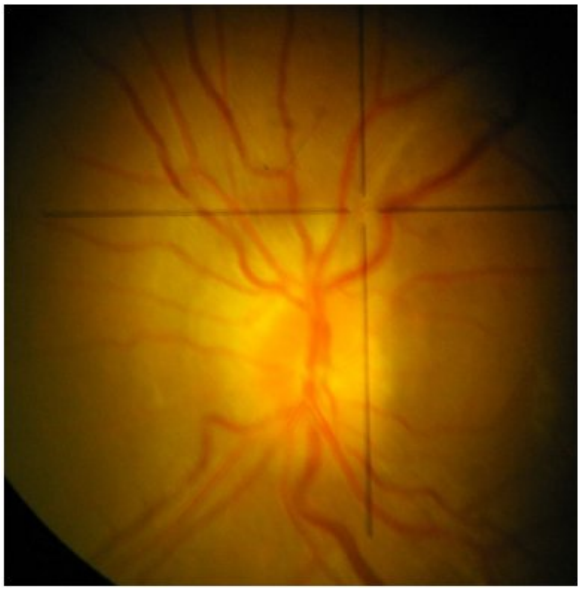
Fundus examination after dilatation of pupil revealed bilateral overt disc edema. [Table/Fig 1] and [Table/Fig 2]



[Table/Fig 1] Fundus photograph of OD showing optic disc edema in a patient with familial Mediterranean fever.

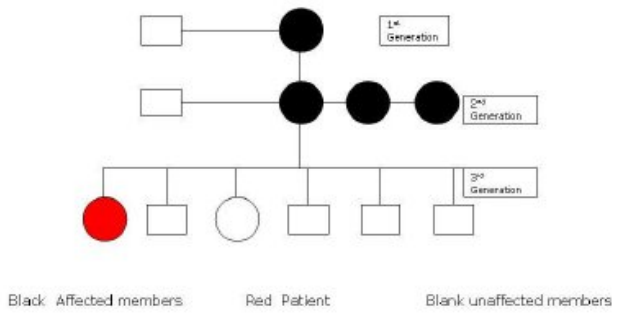
Her BMI was 27.5 kg/M^2. There was no past history of diabetes mellitus or hypertension. She denied any past or present history of headache. She reported recurrent episodes of pain in abdomen & extremities along with fever since childhood. These episodes used to last for two to three days. During an episode, the pain in extremities hampered her day to day activities. She was diagnosed to have familial

Mediterranean fever (FMF); however she was not taking any regular medication. Further interviewing her revealed that episodes of similar nature occurred in other family members in last three generations ([Table/Fig 3]). Interestingly, her grandmother was erroneously operated for appendicitis during an episode of FMF.



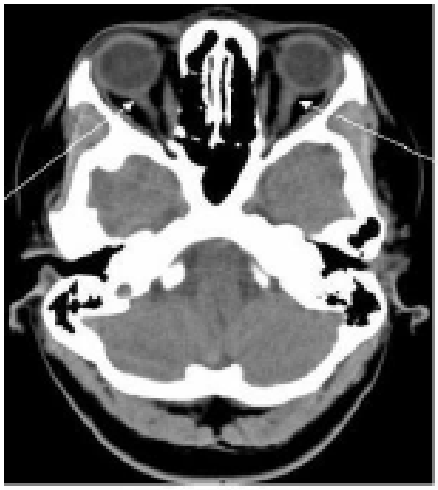
[Table/Fig 2] Fundus photograph of OS showing optic disc edema in a patient with familial Mediterranean fever.

Her Complete blood counts, Erythrocyte sedimentation rate, urine analysis, fasting blood sugar, serum electrolytes, serum calcium, blood urea, serum creatinine, liver function test, TSH, T3 and T4 were within normal limit. Anti-nuclear antibodies were not detectable.



[Table/Fig 3] Pedigree of patient.

Detailed neurological examination of this patient to elucidate her bilateral disc edema was not rewarding. Contrast enhanced computerized tomogram (CT) scan of brain was performed to exclude a mass lesion or sinus thrombosis causing the bilateral disc edema. The only abnormal finding noted was the enlargement of optic nerve sheaths bilaterally ([Table/Fig 4]). CT scan of optic nerve excluded the possibility of buried optic nerve drusen mimicking papilloedema.



[Table/Fig 4] Axial computed tomography demonstrating bilateral enlargement of optic nerve sheath suggestive of raised intracranial pressure.

Despite thorough, repeated history and detailed systemic/neurological examination I was unable to pinpoint any etiological factor that may cause or need further investigation to rule out as a cause, for bilateral disc edema present in my case.

Bilateral disc edema in the clinical settings of normal ocular, neurological and systemic examination along with normal neuro-imaging was thought of probably due to pseudotumor cerebri. Lumbar puncture to document the opening pressure of cerebrospinal fluid was refused by the patient. Treatment was commenced with tablet acetazolamide 250 mg four times a day. Three weeks later, while on tablet acetazolamide, her disc edema was regressing but visual acuity remain same.

Discussion

This patient, suffering from FMF, presented with complaints of blurred vision and bilateral disc edema with no detectable cerebral mass lesion. She did not suffer from headache which is in contrast to the majority (94-99%) of

patients suffering from pseudotumor cerebri. Most of the patients suffering with pseudotumor cerebri are young female, either overweight or have a history of recent weight gain. None of these features were documented in my patient and additionally, she denied taking any medication that could have been responsible for her visual findings. This disease was noted in her previous three generations and, interestingly, all affected members were females.

Pseudotumor cerebri in a patient suffering from FMF was reported by Gokalp et al however their patient presented with headache [9]. Their patient was normal weight female and all her relevant laboratory and radiological parameter were normal. They did not comment about the similar disease in other family members.

Enlarged optic nerve sheath have been reported in various studies in patients with pseudotumor cerebri [10],[11] and it seems to represent a direct consequence of long term increased pressure in CSF spaces [12]. One study correlated the cerebrospinal fluid opening pressure with the diameter of optic nerve [13]. I presume that the enlarged optic nerve sheath in my case represent the radiological evidence of high cerebrospinal fluid pressure.

This case has one limitation. The cerebrospinal fluid pressure, one of the Dandy criteria [14], was not documented in this patient, as she refused for lumbar puncture. It is not possible to say categorically that pseudotumor cerebri was due to FMF. The co-existence of two may be purely coincidental however I believe that bilateral disc edema in this patient was due to FMF and wanted to report this observation with a view to promote awareness and hopefully to stimulate further research to elaborate the relation between pseudotumor cerebri and FMF.

Conclusion

This report highlights that provisional diagnosis of pseudotumor cerebri should be considered for deteriorating vision of recent onset in a patient suffering from familial Mediterranean fever.

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Editorial Comment

Familial Mediterranean Fever (FMF) is a disease of many faces. Almost all the organs are somehow affected. Visual and central nervous system involvement however is not common. In the present case, fundoscopic findings show bilateral papilloedema, and the physical findings propound a possible high intracranial pressure (ICP) which may be diagnosed as Pseudotumor Cerebri (PC) despite lack of a lumbar puncture. The enlargement of optic nerves on CT scan in this case is interesting and can explain the papilloedema following an increased ICP. The enlarged retrobulbar optic nerve most probably exerts anteroposterior compression on the globe evidenced by the posterior scleral flattening associated with idiopathic intracranial hypertension [15]. Madill and Connor's neuroimaging study shows significantly shorter axial length of the eye globes in patients with idiopathic intracranial hypertension compared with control subjects [16]. ICP has been shown to be correlated with intraocular pressure (IOP) [17]. A measurement of the IOP could also be helpful as an easy and non-invasive test in evaluation of this patient. Although, the presumptive diagnosis of PC questions the validity of this paper to some extent, a possible correlation or even coincidence of FMF, visual problems and increased ICP is very interesting, and deserves attention.

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