CASE REPORT

Visual Loss Secondary to Bioccipital Calcifications Associated with Coeliac Disease

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ABSTRACT

The purpose of this paper was to present a case report of permanent visual loss secondary to occipital lobe calcifications in coeliac disease. A 58-year-old grave digger was referred by his work place occupational health and safety department for vision assessment. His past medical history included coeliac disease (CD) diagnosed 20 years previously, as well as an over 40-year history of poor vision that had not been investigated. Examination showed bilaterally decreased visual acuity of <6/60 and a right homonymous inferior quadrantanopia confirmed on automated perimetry. The optic discs appeared normal with no pallor or neuroretinal rim loss. Computed tomography (CT) and magnetic resonance imaging (MRI) scans revealed bilateral occipital calcifications. Diffusion tensor imaging showed reduced anisotropy and difficulty tracing the optic radiations to the occipital lobes. This is the first described case of visual loss secondary to occipital lobe calcifications in coeliac disease.

Keywords: Calcification, coeliac, occipital, visual loss

INTRODUCTION

Coeliac disease is caused by a T-lymphocyte-mediated abnormal immune response to gluten in the small intestine. This inappropriate response causes damage to intestinal mucosa and malabsorption of the fat-soluble vitamins vitamins A, D, and K and folate.¹ The malabsorption consequence can result in various extra-intestinal manifestations.

Coeliac disease, epilepsy, and cerebral calcifications (bilateral cortico-subcortical occipital lobe calcifications) (CEC) is an extra-intestinal manifestation of coeliac disease and is attributed to poor intestinal absorption of folate. Permanent visual loss with the bioccipital calcifications has not been previously reported.

We here report the first case of retrochiasmal visual loss from coeliac disease due to bioccipital calcification. Informed consent was obtained from the patient involved.

CASE REPORT

A 58-year-old grave digger was referred by his work occupational health and safety officer for vision certification. He had been employed for over 30 years in the same place, but a recent job specification update required the use of a computer and ability to operate a ride-on lawn mower, which he was unable to do. He had poor vision for over 40 years that was uninvestigated. His coeliac disease was diagnosed 20

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FIGURE 1 (A) Fundus photograph of both optic discs showing normal optic nerve without evidence of optic atrophy. (B) Visual fields grey scale (top) and pattern deviation showing a right inferior quadrantanopia. (C) Axial CT scan showing bioccipital calcification. (D) MRI T1-weighted image with hypointensity in the occipital region corresponding to the area of calcification. (E) Diffusion tensor imaging of the visual pathway and the optic radiation (OR) on the left is missing.

years previously with a positive bowel biopsy. At the time of diagnosis, he recalled being told that he had vitamin A and vitamin D deficiency. He had been treated with appropriate vitamin supplements and folic acid but was currently not taking any

medications, and his coeliac disease was well controlled with a gluten-free diet. He denied any other significant past medical or family history. In particular, there was no pattern of a hereditary optic neuropathy.

On examination, his best-corrected visual acuity was 6/95 in the right and 6/75 in the left eye. There was no relative afferent pupillary defect. Ishihara colour vision was 3/15 bilaterally. Motility and cranial nerve examination was normal. Slit-lamp examination showed clear anterior segments. The optic nerves were normal (Figure 1A) with no pallor or neuroretinal rim loss. Automated perimetry revealed a right incomplete inferior quadrantanopia (Figure 1B).

A computed tomography (CT) scan revealed bilateral occipital calcification (Figure 1C), which was also revealed on magnetic resonance imaging (MRI) (Figure 1D). There was also associated posterior cerebral atrophy and restricted diffusion. Diffusion tensor imaging (Figure 1E) to visualize the optic pathways was reconstructed and showed reduced anisotropy and missing optic radiations.

The patient's nutritional biochemical markers for vitamins A, D, folate, and B₁₂ were all in the normal range. He also had electroretinography, which was normal, excluding retinal pathology and visual evoked potentials, which revealed normal latency but reduced amplitude bilaterally, suggestive of axonal loss.

DISCUSSION

Permanent visual loss from bi-occipital calcification from any cause has never been reported, and its mechanisms are not completely understood. Bilateral occipital calcification in CEC is thought to be due to coeliac disease (CD)-caused folate deficiency.² In our patient this cause seems unlikely, as he has been taking long-term folate supplementation and has normal serum levels. That said, his vision decline predates his diagnosis of coeliac disease and his treatment (gluten free diet) by at least 20 years. Damage via a nutritional deficiency could have occurred in the 20 years prior to diagnosis, meaning he could have had CD much earlier and been asymptomatic. Even an earlier biopsy in such a patient may have been negative due to "patchy mucosa." Therefore, exposure to gluten and a potential previous nutritional deficiency may be one initial cause for calcifications in such patients.

The findings of vision loss in our patient is likely retrochiasmal given the homonymous quadrantanopia on visual field testing and normal optic nerve appearance without any optic pallor or atrophy. This suggests at minimum that pathogenic progression and/or initiation has occurred via a different mechanism, which we believe to be genetic/immunological. Disorders that can cause occipital calcification and homonymous field defects include Sturge-Weber syndrome and other causes that result in folate deficiency, including treatment with methotrexate and congenital folate malabsorption.^{3,4} Some occipital

benign tumours may have calcification within the lesion, such as a meningioma or teratoma, but the appearance of these are usually within a well-defined lesion and are different radiologically to the diffuse ribbon-like calcification in coeliac disease.

Furthermore, the homonymous field defect and reduced bilateral visual evoked potentials suggesting axonal loss in our patient suggest two more elements that support our hypothesis: (1) a mechanism causing potential neuronal destruction and (2) a retrochiasmal process for visual loss, proven by diffusion tensor imaging being unable to visualize the optic radiations, suggesting occipital calcification.

Visual loss in CD has only once been previously reported.⁵ Visual acuity change in coeliac disease (CD) can result from vitamin A deficiency, but this again is unlikely in our patient, as he did not have night blindness, corneal xerosis, or keratomalacia.⁶ Our patient's fundus examination and full-field electroretinography were normal.⁷ Furthermore, vitamin A optic neuropathy usually causes a caeco-central visual field scotoma, with optic nerve temporal neuroretinal rim pallor from optic atrophy. These features are not present in our patient.

The reason why calcification has a predilection for the occipital lobes in CEC is unknown.⁸ Patients with CEC also typically have epilepsy; however, our patient has no seizure history. We believe this is because there are atypical CEC variants that can exist through linkage disequilibrium⁹ via different combinations of genes close to the HLA-DQ2 region, to which 90% of cases CD and CEC are linked.^{10,11}

We hypothesise that the primary cause in our patient and other sufferers of CEC is neuro-immunological, initiated by gliadin-activated T cells, which cross the blood-brain barrier and release pro-inflammatory cytokines. These cytokines then cause deleterious/toxic effects on central nervous system (CNS) cells and myelin and also deregulate calcium permeability leading to CC.⁸

Prior exposure to gluten may cause enteropathy but this can be asymptomatic, as neurological symptoms can occur without enteropathy. This further supports a neuro-immunological cause for visual decline.⁸ At best, in our patient, prior folate deficiency was only contributory.

Finally, due to the linkage of dysequilibrium and heterogeneity between CEC patients, it is possible that the epilepsy manifestation in a person in CEC may be absent or occur later in life. Only one other reported case of CD causing optic atrophy and progressive visual decline has been reported.⁵

CONCLUSION

This is the first reported case of permanent visual loss from bioccipital calcification from CD and the second

case of visual loss in CD.⁵ The mechanism for vision loss from CD is still largely unknown. We hypothesise that a major cause and the likely cause in our patient, a potential CEC atypical variant, was genetic and immunological, leading to inflammatory changes resulting in CC and damage to retrochiasmal CNS tissue.

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