Macular pattern dystrophy in MIDD: Long-term follow-up


Abstract

A case of maternally inherited diabetes and deafness (MIDD)-associated macular pattern dystrophy with a 15-year follow-up is reported. On initial examination at age 37, visual acuity was normal, but chorioretinal atrophy at the posterior pole was already present in both eyes. At age 52, visual acuity remained normal in the right eye and was only slightly decreased in the left eye despite notable extension of the areas of chorioretinal atrophy in that eye. No evidence of diabetic retinopathy was present at any time. This case shows that visual acuity can remain stable in the long term despite extensive lesions of macular pattern dystrophy.

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Maternally inherited diabetes and deafness (MIDD), due to a 3243 mtDNA point deletion, is characterized by the association at various degrees of diabetes, neurosensory deafness and a lesion of the retinal pigment epithelium called “macular pattern dystrophy” [1]. The latter is present in up to 85% of MIDD patients of Caucasian origin [2], and is strongly evocative of MIDD in the context of diabetes. We present the long-term evolution of an MIDD patient who presented with increasing severity of pattern dystrophy, but relative preservation of visual acuity, and no appearance of diabetic retinopathy.

1. Case report

Mr. B.J. was diagnosed with diabetes at age 30 with the onset of symptomatic hyperglycaemia (3 g/L) and weight loss (body mass index [BMI] 19 kg/m²). The association with progressive hearing loss led to the presumptive diagnosis of MIDD, which was confirmed by the presence of the 3243 mtDNA mutation. Family history revealed gestational diabetes and premature death in the case of his mother and non-insulin-dependent
diabetes and deafness in a sister (who was later found to be also positive for the 3243 mtDNA mutation). Diabetes was treated with antidiabetic oral agents for 15 years, then with insulin by a twice-daily injection treatment regimen with premix insulin. Coenzyme Q10 treatment (ubidecarenone, 150 mg/day) was begun at age 45. At age 37 (1992), macular pattern dystrophy was identified on retinal photographs and angiography (Fig. 1). No diabetic retinopathy was present. Visual acuity was normal at that time in both eyes. Fifteen years later (and 22 years of diabetes), urinary albumin excretion was 128 mg/24 h and creatinine plasma levels were 91 umol/L. At that time, the patient complained of vision loss in the left eye. The best corrected visual acuity was 20/25 in the right eye and 20/40 in the left eye. Fundus examination revealed extension of chorioretinal atrophy, especially in the left eye (Fig. 2). There was no diabetic retinopathy at any time.

2. Discussion

In 1995 [1], we reported on two probands from two different families with MIDD bilateral macular pattern dystrophy of the retinal pigment epithelium, with annular and radiating patterns of pigmented lesions surrounding the macula. This association was then confirmed in further reports [3–5]. Prospective studies by the GEDIAM group [6] showed a high prevalence of macular pattern dystrophy (42/49, 86%), but little visual impairment (43/49 had normal visual acuity). Moreover, among the six patients with visual impairment and macular dystrophy, two had visual loss for unrelated reasons. No correlation was found between the severity of macular dystrophy and age or diabetes duration, although atrophic forms were mostly seen in older patients. Comparison with non-MIDD diabetic patients has shown that MIDD patients have a lower incidence of diabetic retinopathy. The presence of macular dystrophy...
was also described in a large Japanese study that included 113 patients with MIDD [7]. Only 13.4% had macular pattern dystrophy with small pigmented lesions localised on the macula (grade 1). None had visual loss. Of these patients, 55% presented with diabetic retinopathy, a higher prevalence than the 8.7% rate observed in French MIDD patients [2], confirming the inverse relationship between macular dystrophy and diabetic retinopathy. Yet, the cause of the difference in the incidence of macular dystrophy remains to be determined.

Macular pattern dystrophy associated with MIDD usually appears at around age 25 years and begins with mildly pigmented lesions of retinopathy localised to the posterior pole. It may then progress to more severe cases with major chorioretinal atrophy of the posterior pole. The visual prognosis is usually relatively good except when chorioretinal atrophy reaches the fovea, thus leading to visual loss as observed in this patient.

In conclusion, macular pattern dystrophy, which is highly prevalent in MIDD patients of Caucasian origin, is asymptomatic in most cases, even where the lesions are widespread. Our case also illustrates the differences between extensive retinal atrophy and complete absence of diabetic retinopathy.

Conflicts of Interest

None in the field of the study.

References