A rare ophthalmologic disorder: Gyrate atrophy with sparse hair

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Gyrate atrophy (GA) is a rare, progressive metabolic choroid and retinal degeneration that results from a deficiency of the pyridoxal phosphate-dependent mitochondrial matrix enzyme ornithine aminotransferase. Here, we report the case of a 40-year-old woman who presented with a gradual decline in visual acuity since puberty, along with a history of high myopia and cataract surgery. She was admitted to the Dermatology Clinic with chief complaints of sparse hair on her scalp, eyelids, eyebrows and other areas of the body for the previous 5 years. Physical examination showed that scalp hair along with hair from other parts of her body were fine, straight, and sparse with areas of non-well defined alopecia. Hyperornithinemia was documented during laboratory evaluation of the patient.

Keywords: gyrate atrophy, ornithine, ophthalmology, alopecia

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INTRODUCTION

Gyrate atrophy (GA) is a rare, progressive metabolic choroid and retinal degeneration that results from a deficiency of the pyridoxal phosphate-dependent mitochondrial matrix enzyme ornithine aminotransferase mapped on chromosome 10q26. Hyperornithinemia and ornithinuria have been recognized as the biochemical marker for this disorder in 1973 [1].The incidence is highest in Finland, with an estimated frequency of about 1 in 50000 individuals and an estimated frequency for heterozygotes of 1 in 110 individuals [2]. The major clinical problem in these patients is a slowly progressive loss of vision that leads to blindness, usually by the fifth decade of life. Aside from visual impairment, patients with GA are for the most part asymptomatic [3]. Some patients may rarely experience hair loss and muscle weakness. Here, we present the case of a 40-year-old woman with hyperornithinemia and sparse scalp and body hair [4].

CASE REPORT

A 40-year-old married woman presented with a gradual decline in visual acuity since puberty. She had a history of high myopia and cataract surgery with an intraocular lens implant 2 years ago. She came to our clinic with chief complaints of sparse hair on her scalp, eyelids, eyebrows and other areas from 5 years ago (Figure 1). Physical examination showed that her head, other body sites, and genital hairs were fine, straight, and sparse with areas of non-well defined alopecia. Microscopic examination revealed that the frontal, occipital, and pubic hairs were a light brown color without any hair shaft abnormality. Biopsy of the scalp was non-specific and revealed some terminal and telogenic hair follicles placed normal hair follicle with fibrosis, neovascularization, lymphocyte infiltration, along with mild exocytosis in the epidermis. Incontinence of melanin pigment was also observed (Figure 2).

The patient is the result of a consanguineous marriage. The same disorder is present in her father, one sister, and both brothers; all have complaints of decreased visual acuity since early childhood and high levels of plasma ornithine (>900 nmol/ml), approximately 10 times the normal value (10-163 nmol/ml). Her father is 84 years old and blind due to GA of the retina and choroid. Her sister is

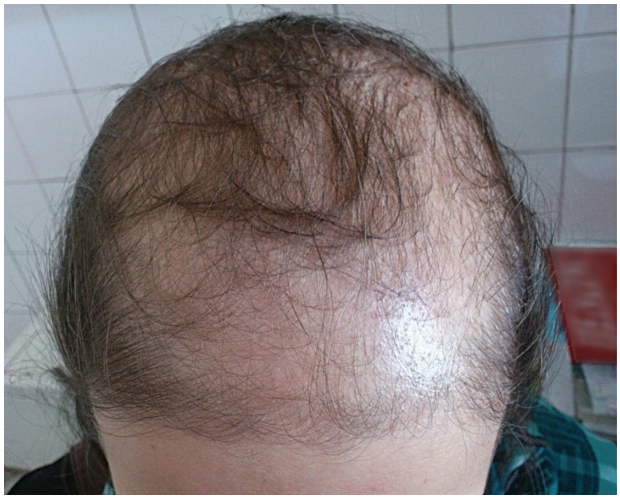

Figure 1. Patient with gyrate atrophy (GA) disease and sparse scalp hair.

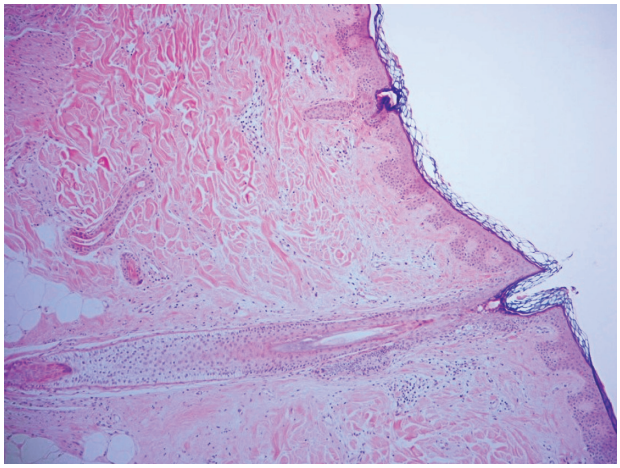

Figure 2. Scalp biopsy reveals non-specific changes (H & E, 40×).

30 years old, one brother is 35 and the other is 48 years old without any history of sparse hair on the scalp and body.

The patient has two sons, 7 and 4 years of age. Both deny any problems. She has received vitamin B6 supplementation for her disease since several years ago, without any improvements in her vision and hair abnormality.

DISCUSSION

Gyrate atrophy of the choroid and retina, or GA, is an inherited disorder characterized by progressive vision loss. People with this disorder have an ongoing loss of cells in the retina, which is the specialized light-sensitive tissue that lines the back of the eye, and in a nearby tissue layer called the choroid [5]. During childhood, they begin to experience myopia and loss of side vision. Over time, their field of vision continues to narrow, resulting in tunnel vision. Many people with GA also develop cataracts. These progressive vision changes lead to blindness by about the age of 50 years; additional clinical symptom have been either minimal or absent [6].

Montagna and Parakkal *et al.*, for the first time, conducted a study about the hair of ten patients with GA and hyperornithinemia. They found that all patients had peculiar, fine, straight hair. The scalp hair was sparse and resulted in areas of alopecia. Gross examination of the hair revealed an absence of abnormal breakage, bending, or ring formation. The scalp hair from all ten GA patients had a similar clinical appearance in both texture and sparseness. When these patients were examined further, the majority of both scalp and pubic hair showed a strikingly abnormal morphology with a dark central core apparent on both light and electron microscopy [7]. The dark core areas seen in the central zone of the hair were not a deposit of an abnormal substance but appeared to be due to the unusual refractive properties of loosely formed macrofilaments. It has been hypothesized that this characteristic microscopic appearance may be related to an abnormal structural substance deposited in spaces ordinarily filled with macrofibrils [7,8]. Spaces between the macrofibrils have been described to contain mucopolysaccharide-rich substances which take a longer time in the dehydration process of hair maturation. The dark cores in both pili annulati and GA may be related to the refractile properties of loosely formed macrofilaments. In the case of pili-annulati, the hair shaft contains periodically occurring clusters of air-filled cavities, previously filled with a substance [9-10]. In GA, the spaces still appear to contain a structure-less electron lucent, compact substance. Ornithine aminotransferase deficiency may contribute to this defect in hair maturation. The occurrence of this finding is significantly greater than 85% of the hair from patients [4]. Although the current study patient had diffuse alopecia we did not observe such abnormalities under light or polarized microscopy. Multiple samples would be needed to locate this finding, however the patient refused the recommendation for repeated sampling. She has received treatment with vitamin B6. However,

it is debatable whether pyridoxine can decrease level of ornithine and affect retinal atrophy [5]. A review by Hayasaka *et al*. has stated that out of the over 150 biochemically documented cases of GA, approximately one third were from Finland and only seven (less than 5%) responded to vitamin B6 dietary supplementation [11]. We have not located any study about the effect of vitamin B6 on hair abnormality in patients with GA. Our case is the first report of alopecia in GA for a dermatologist. Although GA is an ophthalmologic condition, dermatologists should be aware of this disease and resultant hair abnormality. We did not find GA or ornithine aminotransferase deficiency as a result of hair shaft abnormality in a dermatology text book. It would be of interest to confirm this in the future by examining additional patients with GA.

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