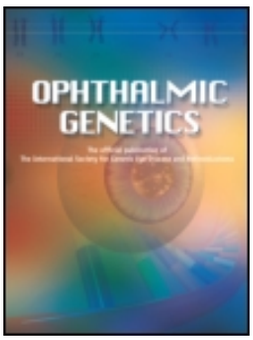

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Eric Souied, Pierre Amalric, Marie-Liesse Chauvet, Christine Chevallier, Phuc Le Hoang, Arnold Munnich & Josseline Kaplan

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Unusual association of juvenile macular dystrophy with congenital hypotrichosis: occurrence in two siblings suggesting autosomal recessive inheritance

Eric Souied[1]
Pierre Amalric[2]
Marie-Liesse Chauvet[1]
Christine Chevallier[3]
Phuc Le Hoang[4]
Arnold Munnich[1]
Josseline Kaplan[1]

[1]Service de Génétique et Unité de Recherches sur les Handicaps Génétiques de l'Enfant, INSERM U. 393, Hôpital des Enfants Malades, Paris, France
[2]Consulation d'Ophtalmologie, Albi, France
[3]Consulation d'Ophtalmologie. Gien, France
[4]Consulation d'Ophtalmologie, Hôpital Pitié-Salpétrière, Paris, France

Abstract A familial association between juvenile macular dystrophy and congenital hypotrichosis is described in two siblings aged 25 and 23 years. We put forward arguments for locating the retinal alteration at the level of the retinal pigment epithelium and suggest that the hair disorder could be a Marie-Unna type hypotrichosis. This association is transmitted as an autosomal recessive condition.

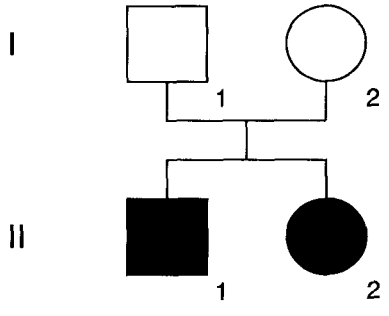
Key words Hypotrichosis; macular dystrophy; hereditary

Correspondence to: Josseline Kaplan, Service de Génétique et Unité de Recherches sur les Handicaps Génétiques de l'Enfant, INSERM U. 393, Hôpital des Enfants Malades, Paris, France.

Introduction A familial association between juvenile macular degeneration and congenital hypotrichosis was described for the first time in 1935 by Wagner[1] in two sisters. A second report was published by Yasakura et al.[2] in 1967, a third by Johnston et al.[3] in 1973, and a fourth by Kroll[4] in 1981. Here, we report the association between congenital hypotrichosis manifesting as generalized loss of hair and juvenile macular degeneration characterized by progressive dystrophy of the retinal pigment epithelial (RPE) layer in the posterior pole in two siblings of a Portuguese family. The healthy parents were born in the same small village, giving further support for autosomal recessive inheritance.

Case reports The proband (II1, Fig. 1) was the eldest son of Portuguese non-consanguineous parents. Since birth, he has had abnormally sparse hair (Fig. 2) and at the age of 25 years had still never cut his hair. Nevertheless, he had no other symptom of ectodermal dysplasia, including normal teeth and

Fig. 1. The family pedigree.



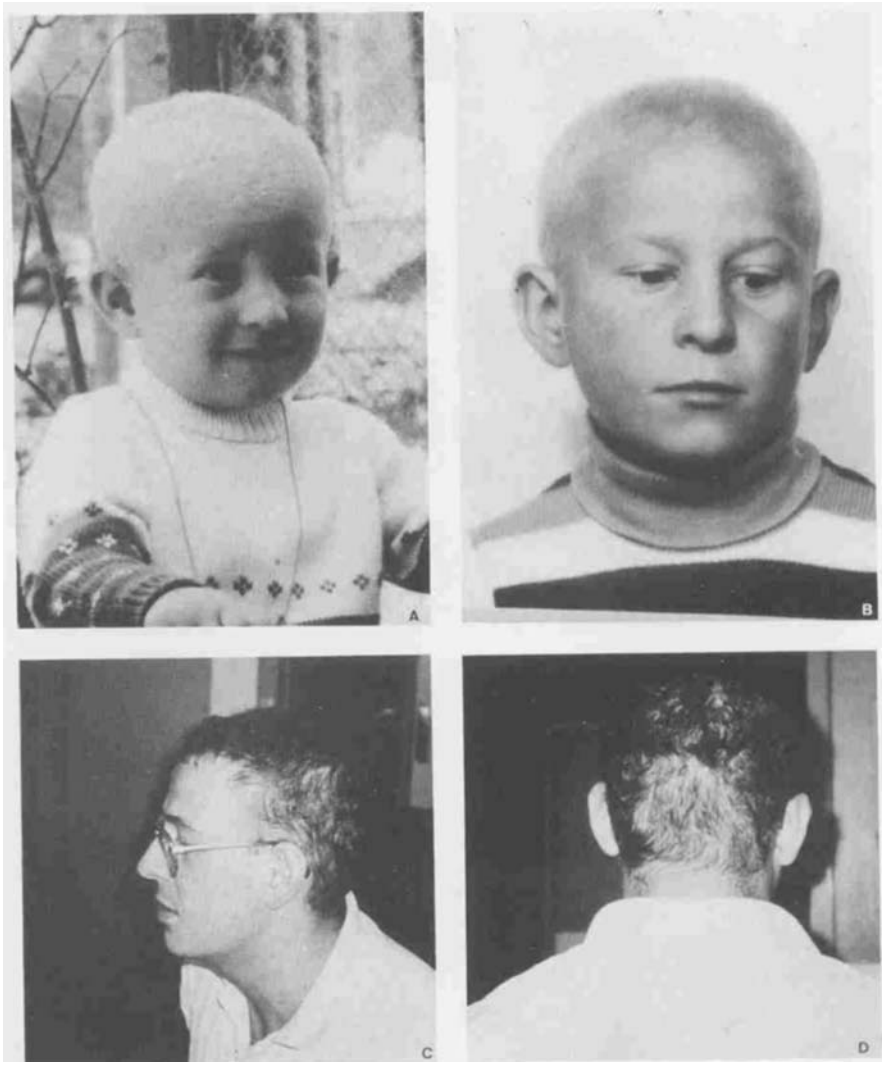


Fig. 2. Probands's (II1) aspect at different ages: A) at the age of 4 years, B) at the age of 12 years, C) and D) at the age of 25 years, note the abnormally sparse hair.

nails. When he was 18 years old, he complained of progressive reading difficulties, and was examined by an ophthalmologist. At that time, examination revealed reduced visual acuity and macular pigmentary degenerative changes. At the age of 25 years, his visual acuity was about 20/200 in both eyes and he had very reduced reading ability. He had a bilateral central scotoma on visual field recording and a mild loss of color perception. Fundus examination showed bilateral degenerative changes within the macular region (Fig. 4). Fluorescein angiography (Fig. 5) showed a macular central geographic atrophy with some peripapillary involvement, pigment epithelial atrophy associated with pigment clumping, and some remaining areas of normal pigment epithelium. Late mottling of the choroid is visible on the temporal border. The electroretinogram (ERG) was almost normal for both scotopic and photopic responses. In contrast, the electro-oculogram (EOG) displayed bilateral severely reduced responses with an Arden ratio of 0.5 for the right eye and 0.3 for the left eye (normal ratio 1.8).[5]

The proband's sister (II2, Fig. 1) has the same hair abnormality as her brother (Fig. 3). At the age of 23 years, she is very embarrassed by her appearance and usually wears a wig. Although, she never complained of visual diffi-

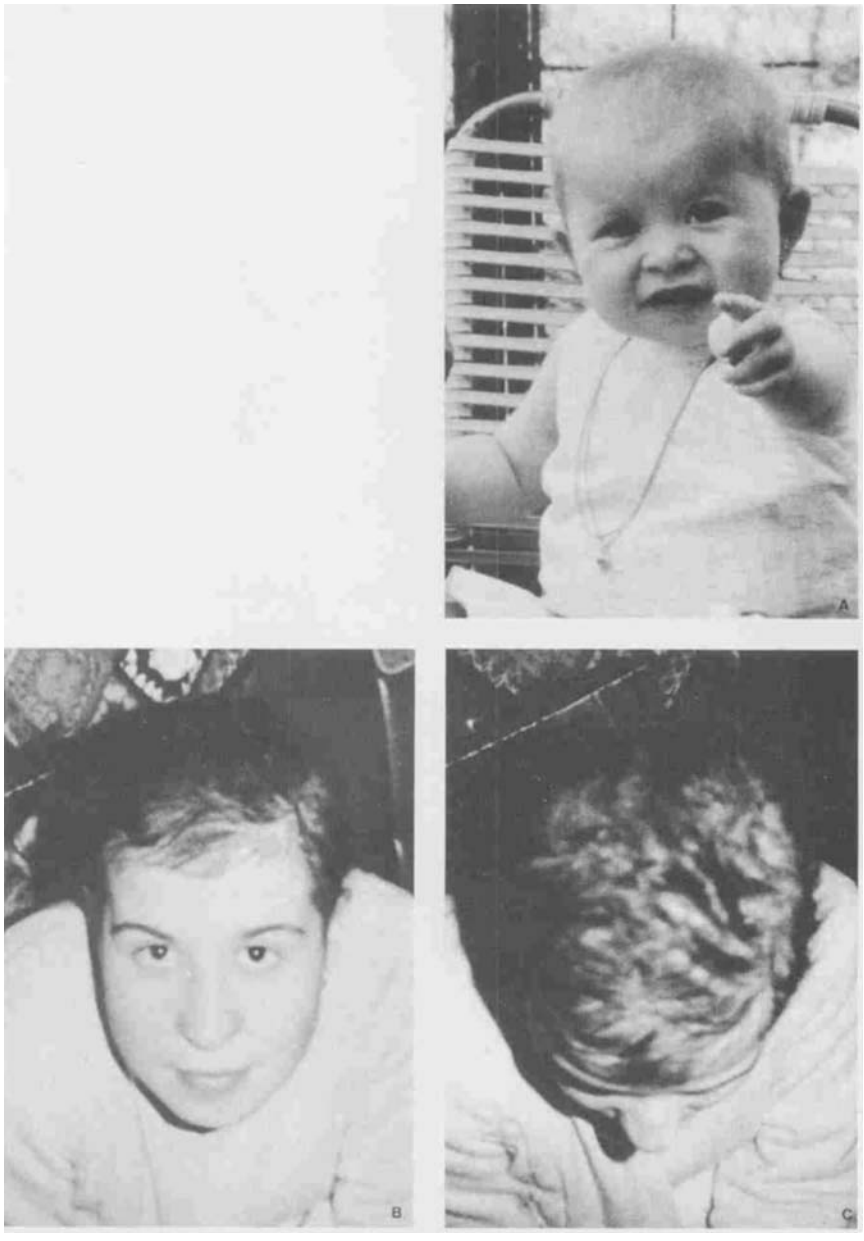


Fig. 3. Probands' sister (II2) A) at the age of 2 years, B) and C) at the age of 23 years. She has never cut her hair since birth.

culties, she was examined by the same ophthalmologist as her brother after he was found to have retinal changes. Examination revealed the same macular dystrophy of the pigment layer of the posterior pole. Unfortunately, she has refused further investigations.

Discussion In this paper, we report the observation of an unusual association between juveline macular dystrophy and congenital hypotrichosis. Ophthalmoscopy and fluorescein angiography located the alteration at the level of the RPE. This was confirmed by the normal ERG and the abnormal EOG.

Few disorders are known to have this type of dissociation between normal ERG and abnormal EOG and until now it was said to be the hallmark of vitelliform macular dystrophy (Best disease).[6,7] Nevertheless, this dissociation is

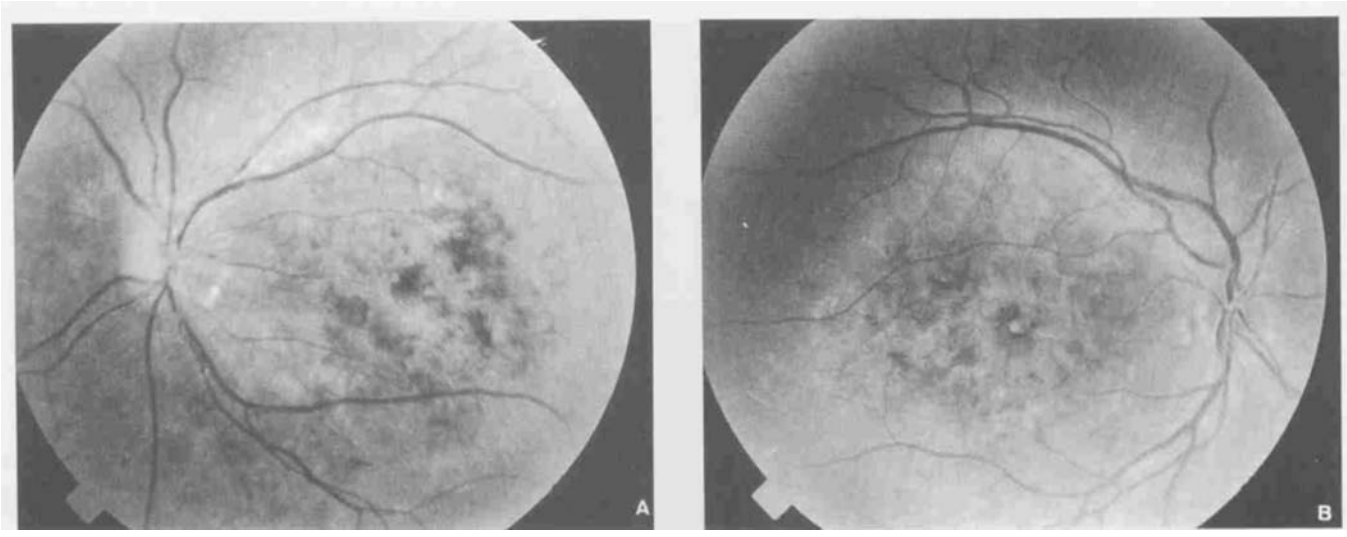


Fig. 4. Proband's fundus, note the degenerative changes within the macular region. A) left eye, B) right eye.

also seen in butterfly-shaped dystrophy[8] and in the pattern dystrophy of Marmor and Byers.[9] The RPE changes observed in these three disorders, however, are clearly different from those observed in our patients. In addition, these affections are inherited as an autosomal dominant trait, while in our family and in those reported by Wagner[1] and Yasukura et al.[2] the most probable mode of transmission is autosomal recessive.

As far as hair abnormalities are concerned, hypotrichosis occurs in numerous hereditary disorders. In some of them, a well-recognized abnormality of the hair is present, such as pili torti or monilethrix. A clinical subtype of hypotrichosis without any distinctive abnormality is often defined as the Marie-Unna type[10,11] and is characterized by a generalized loss of hair. This is the diagnosis that can be proposed for this family. Usually, Marie-Unna hypotrichosis is not reported to be associated with retinal degeneration. However, one recent publication reported hereditary hypotrichosis (Marie-Unna type) associated with Stargardt's maculopathy.[12] In that report, the two affected siblings displayed a very early onset of bilateral loss of vision and macular

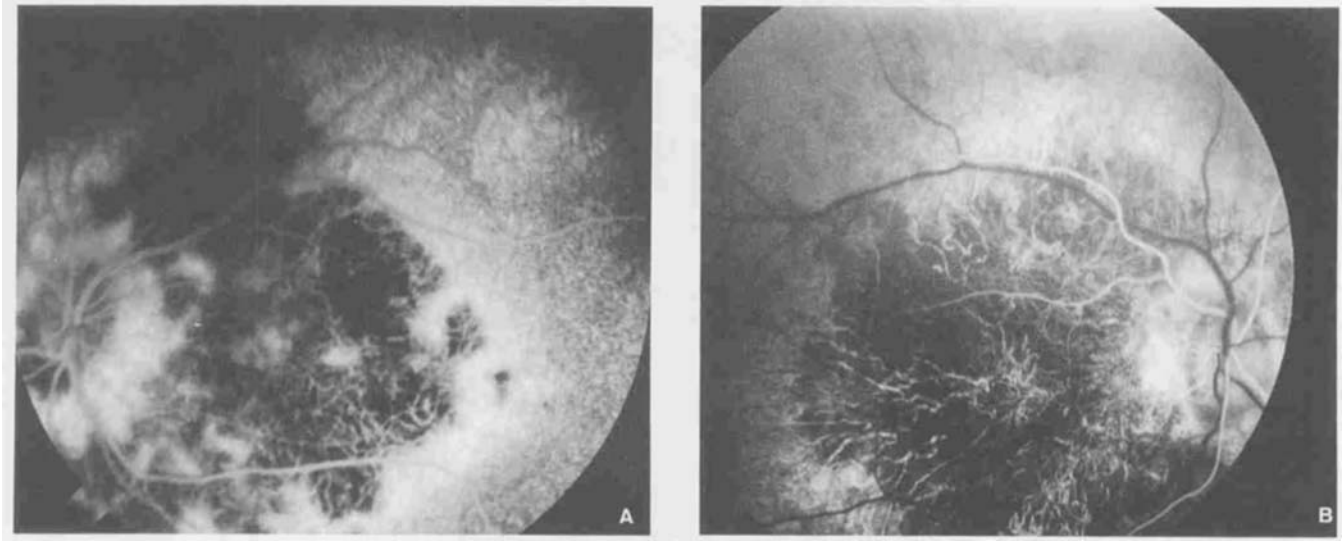


Fig. 5. Proband's fluorescein angiography showing RPE atrophy of the posterior pole, with choroidal vessels visible because of this atrophy. A) left eye, B) right eye.

changes (6 years and 4 years, respectively) Their condition thus appears to be different from that of our patients.

In summary, we describe the fifth instance, to our knowledge, of juvenile macular dystrophy associated with congenital hypotrichosis and put forward arguments for a more precise classification of these conditions. The retinal disorder is most probably a macular RPE dystrophy and the hair disorder a Marie-Unna type hypotrichosis. This association was transmitted as an autosomal recessive condition in the presently reported family.

References

1 Wagner H. Makulaaffektion vergesellschaftet mit Haarabnorma-lität von anugotypus, beide vielleicht angeboren bei zwei Geschwistern. Albrecht von Graefes Arch Klin Exp Ophthalmol 1935; 134: 71.

2 Yasukura K, Mizuta S, Maedak S. Macular degeneration in siblings with unusual hairgrowth. Jpn J Clin Ophthalmol 1967; 21: 495.

3 Johnston SS, Darragh J, Nevin NC. Hereditary macular degeneration in three generations. Br J Ophthalmol 1973; 57: 578-583.

4 Kroll P. Beidseitige kongenitale Pigmentblattdystrophie des hinteren Augenpols bei gleichzeitiger Hypotrichosis congenita totalis. Klin Monatsbl Augenheilkd 1981; 178: 118-120.

5 Arden GB, Barrada A, Kelsey JH. New clinical test of retinal function based on the standing potential of the eye. Br J Ophthalmol 1962; 46: 449-467.

6 Deutman AF. The hereditary dystrophies of the posterior pole of the eye. Van Gorcum, Assen, Netherlands, 1971.

7 Mohler CW, Fine SL. Long term evaluation of patients with Best's vitelliform dystrophy. Ophthalmology 1981; 88: 688-691.

8 Deutman AF, Van Blommestein JDA, Henkes HE et al. Butterfly-shaped pigment dystrophy of the fovea. Arch Ophthalmol 1970; 83: 558-569.

9 Marmor MF, Byers B. Pattern dystrophy of the pigment epithelium. Am J Ophthalmol 1977; 84: 32.

10 Unna M. Über Hypotrichosis congenita hereditaria. Dermatol Wochenschr 1925; 81: 1167-1178.

11 Stevanovic DV. Hereditary hypotrichosis congenita (Marie Unna type). Br J Dermatol 1970; 83: 331-337.

12 Marren P, Wilson C, Dawber RPR, Walshe MM. Hereditary hypotrichosis (Marie-Unna type) and juvenile macular degeneration (Stargardt's maculopathy). Clin Exp Dermatol 1992; 17: 189-191.