

Macular Dystrophy in Deafmutism

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Many reports have been published about the association of retinopathy and deafmutism.

In 1960, Amalric described a syndrome, characterized by macular dystrophy and congenital deafmutism. He found this association 15 times among 200 deafmutes and he thought that it is hereditary. Diallinas (1959) had already noticed a pigmentary alteration of the fundus in 2 cases of deafmutism.

In this macular dystrophy pigment displacement is seen in the macular area. Black spots of different size are disseminated irregularly over a background of pigment dust. Between the pigment spots small whitish or yellow points can sometimes be seen. Visual function is good. There is no hemeralopia and the electroretinogram is normal.

On the other hand, association between true progressive pigmentary retinopathy, leading to blindness, and acquired deafness is since long known as Usher's syndrome, which is seen in 25% of the cases of retinitis pigmentosa. The latter exists in 4% of the cases of deafmutism.

Finally, a pseudo-retinopathy of the pigmentary type (pseudo-retinitis pigmentosa), due to rubella embryopathy, is often also associated with deafness. This retinopathy is generally spread out over the entire retina, but affects mainly the posterior pole. The retinal bloodvessels and the disc are normal. The condition is stationary. The central and the peripheral vision are good. The electroretinogram is generally normal or subnormal.

Observations of rubellar retinopathy, less frequent than congenital cataract, have been published by Gregg (1941), Swann *et al.* (1943), Ellett (1945), Terry (1945), Long and Danielson (1945), Emerson (1959) etc.

Franceschetti and Bourquin (1946) reported 6 cases of rubellar retinopathy, in 3 of whom deafness was also present.

Marks (1946) examined 38 children born deaf after a maternal rubellar infection during pregnancy. In 17 of them he found an abnormal pigmentation of the fundus.

For Amalric *et al.* (1963) the association of macular dystrophy and deafmutism is a hereditary syndrome:

1. They observed a female with this syndrome and found, among the ascendants, 3 cases of deafmutism.

2. They observed also a mother and her 2 daughters; all three had macular dystrophy, deafmutism, nephropathy and microcornea.

Nevertheless, among 30 cases of Amalric's syndrome, Remky *et al.* (1963) found only 4 hereditary cases.

So, exception made for the pigmentary retinopathy with deafness, which is a

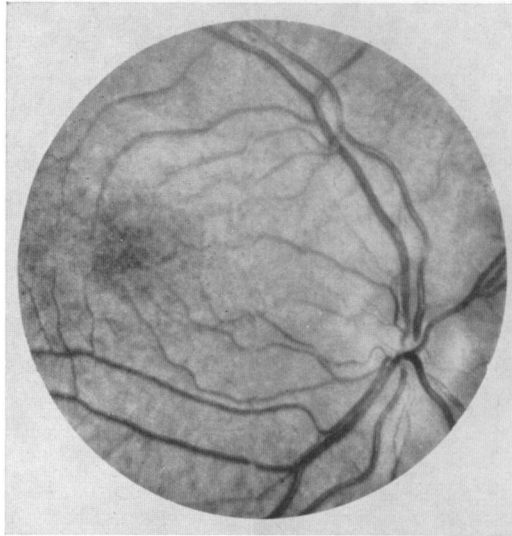


Fig. 1. Macular dystrophy with deafmutism (*pers. obs.*)

true chorio-retinal heredo-degeneration (Usher's syndrome), there are two different points of view concerning the association of retinal alterations with deafmutism: it is either a hereditary syndrome or a manifestation of rubellar embryopathy.

In order to elucidate this problem we have examined systematically 1065 deaf-mutes, i. e. 597 men and 468 women.

Family and personal histories were recorded in detail in order to have all the informations regarding maternal infections during pregnancy and infectious diseases during early childhood and also in order to establish a reliable family pedigree.

The ophthalmological examination of these 1065 children comprised: ophthalmoscopy and examination of the visual functions (central vision, visual field, dark-adaptation and, when possible, electroretinography).

The following results were obtained.

We found 7 cases of true retinitis pigmentosa (0.6%) and 76 cases of pigmentary macular dystrophy of Amalric's type (7%), i. e. 38 men and 38 women.

In all the cases of Amalric's dystrophy the vision was between 0.8 and 1.0, except in 1 case, where it was 0.6 and in a few cases with other ocular anomalies. The visual field was normal.

Among the 76 cases of Amalric's dystrophy 11 (14%) were hereditary and 65 (86%) of unknown (27 or 41%) or peristatic (38 or 59%) origin.

Among the 38 cases of known etiology, 23 (60%) were due to a maternal infection during pregnancy and 15 (40%) to an infection during early childhood.

The maternal infections were distributed as follows: 13 cases of rubella, 7 cases

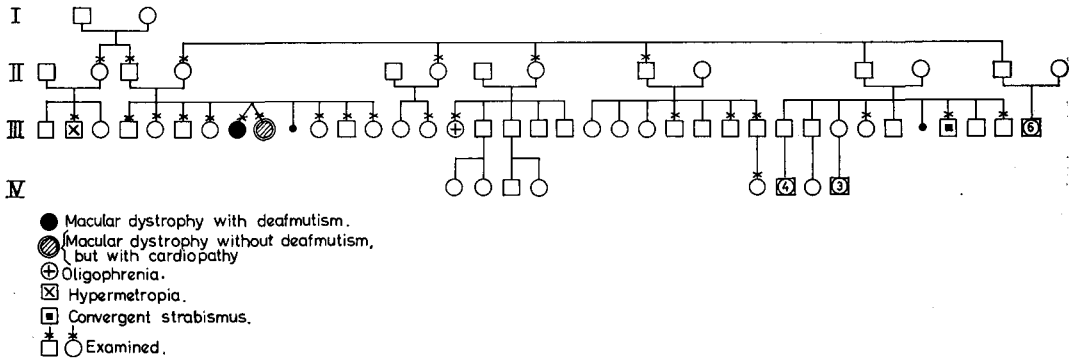


Fig. 2. Pedigree of case I

of viral infection with skinrash (among these the mother of a pair of twins) and 3 cases of possible viral infections.

The infections of early childhood comprised 4 cases of otitis, 5 cases of meningo-encephalitis, 1 case of parotitis and 5 cases of seizures.

These 76 cases of macular dystrophy with deafness were exceptionally associated with other ocular or systemic anomalies. Among the ocular anomalies we have to mention 1 case of unilateral congenital cataract with microphthalmia, 1 case of severe myopia, 2 cases of strabismus with high hypermetropia, 1 case of coloboma of the choroid and eyelid nystagmus and 1 case of microphthalmia. Among the systemic anomalies we noted 1 case of cleft palate and 1 case of cardiopathy.

The study of our cases of macular dystrophy, associated with nonhereditary deafmutism, shows that the anomaly is very often a manifestation of a viral embryopathy. This point of view has to be taken into consideration even for the hereditary cases, at least for the macular dystrophy, because the history of viral infection is here also very important.

We can illustrate this with the following observations in two pairs of twins.

Case I

DZ female twins (Fig. 2), aged 17 (L. and N.), who have anomalies of the sex-chromatin. L. is deafmute; she had, 15 days after birth, a pleuritis with an otitis at the left side. N. is not deafmute, but presents a cardiopathy, which is a frequent sign of embryopathy. Both of them have a pigmentary macular dystrophy, bilateral

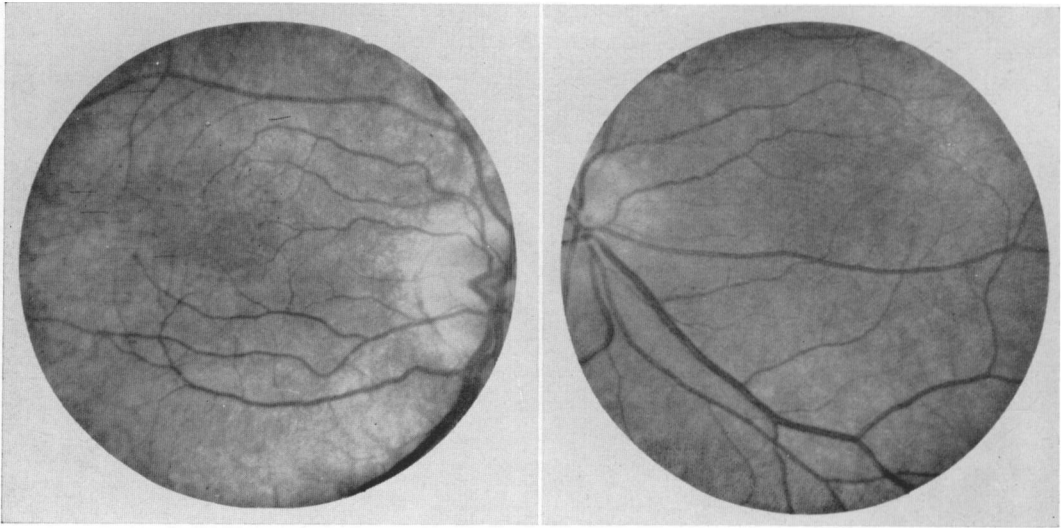


Fig. 3. Bilateral macular dystrophy in twin L. (case I)

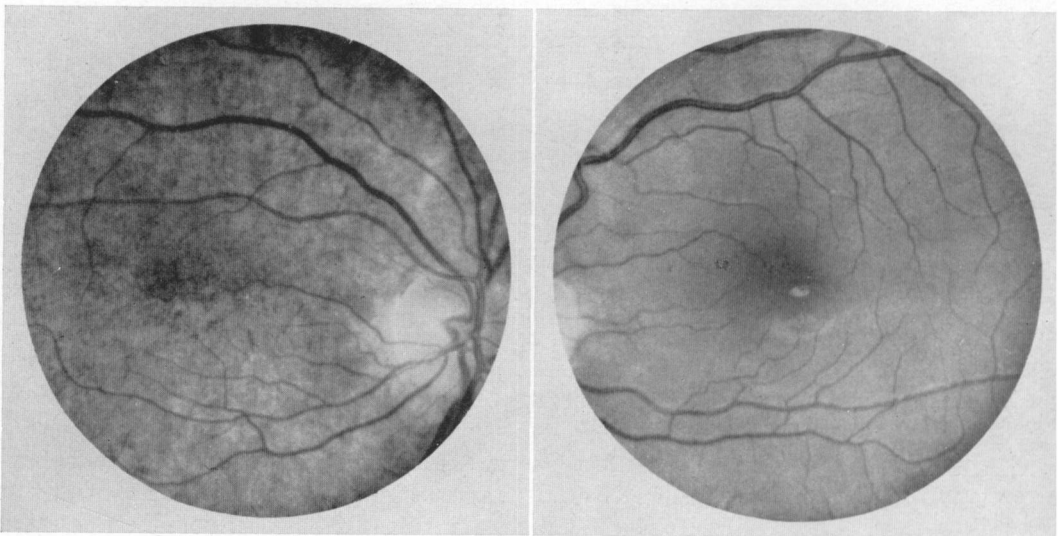


Fig. 4. Unilateral macular dystrophy (R. E.) in twin N. (case I)

in one of them, unilateral in the other. The visual functions (vision, visual field, dark-adaptation, colour vision, electroretinogram and critical fusion frequency) are normal in both.

There is no consanguinity of the parents. The mother had a viral infection with skin-rash during pregnancy. The twins are the 5th and the 6th of a sibship of 9. No auditory nor severe ocular anomalies are seen in the 59 other members of the family. There is only 1 case of high hypermetropia, 1 case of convergent strabismus and 1 case of oligophrenia.

Case II

DZ twins (Fig. 5), aged 18, a boy and a girl. The girl is deafmute. The boy has a keratoconus. Both twins show a pigmentary macular dystrophy. In the girl we see a diffuse dustlike pigmentation of the retina, especially in the macular region,

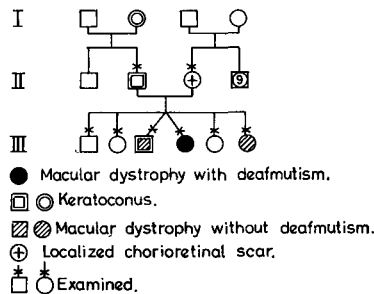


Fig. 5. Pedigree of case II

but also in the periphery, where some atrophic patches can be observed. In the boy we find the same diffuse dustlike pigmentation of the retina; in the macular region 3 atrophic patches with pigmented borders are seen. Both twins have a normal vision and a normal visual field.

The twins are the 3rd and the 4th of a sibship of 6. They had measles at the age of 3 weeks. A younger sister shows also a dustlike pigmentation of the macular region, but she is not deafmute. The other family members have a normal fundus. There are two other cases of keratoconus in paternal ascendants. There is no consanguinity of the parents. The mother was not ill during pregnancy. She shows, in the right eye, at the level of the supero-nasal artery, at 4 disc diameters from the optic nerve, an atrophic chorioretinal patch with pigmented borders.

Thus, it has now been proven that in deafmutes there can exist a pigmentary macular dystrophy, which is different of true pigmentary retinopathy (retinitis pigmentosa) and which is besides characterized by normal visual functions (Tab. 1). This macular dystrophy is found in 7% of the cases.

Tab. 1. Pigmentary macular dystrophy of Amalric's type in deafmutism

Author	Deafmutism N. of cases	Retinitis pigmentosa N. of cases	Macular dystrophy N. of cases
Marks (1946)	38	—	17
Emerson (1958)	7	2	5
Diallinas (1959)	18	1	2
Amalric (1960)	200	1	15
Wuillez (1960)	151	—	9
Grimaud <i>et al.</i> (1962)	179	—	22
Remky <i>et al.</i> (1963)	440	—	30
Amalric and Bessou (1963)	30	2	4
Cernea <i>et al.</i> (1965)	206	2	3
Pers. obs.	1065	7	76
Total	2334	15 (0.6%)	183 (7%)

This association of macular dystrophy and deafmutism, does it really constitute a syndrome? And if so, is it hereditary?

Or can the macular dystrophy exist without associated deafmutism?

Or is it an abortive manifestation of a more complex malformative condition and particularly of a rubellar embryopathy?

And finally, is this macular dystrophy always congenital?

Although the association of deafmutism and macular dystrophy cannot be coincidental, it has to be admitted that heredity does not seem to play a role in the majority of the cases:

1. The cases, which are manifestly hereditary, are exceptional (not more than 14%).

2. Even in the cases, where heredity is an etiologic factor for the deafmutism, a history of pre- or postnatal viral infection indicates the possible intervention of an exogenous factor.

3. The study of our two pairs of twins has failed to prove the hereditary nature of the associated lesions. In the first pair both show a macular dystrophy, but only one is deafmute. This had a neonatal pleuritis with otitis and the other presents a cardiopathy. In the second pair, both show also a macular dystrophy, but only one is deafmute, whereas the other has a keratoconus.

Macular dystrophy can thus exist without associated deafmutism, as has been observed in one member of two DZ twin pairs, of which the other presented deafmutism, and in a female belonging to the same sibship as the second twin pair.

On the other hand, macular dystrophy can certainly constitute an abortive manifestation of a viral or rubellar embryopathy:

1. Marks (1946) observed this dystrophy in 50% of the patients congenitally deafmute by maternal rubellar infection and noted that the other congenitally deafmutes did not show macular dystrophy.
2. We have detected a maternal viral infection in at least 35% of our cases.

Finally, it seems that the macular dystrophy is not always congenital (Diallinas, 1959) — and this is also the case for the deafness — and that it might be due to infectious diseases of the early childhood (measles, epidemic parotitis, vaccinia encephalitis etc.).

In short, most of the cases of Amalric's macular dystrophy associated with deafmutism seem to be due to a maternal rubella during pregnancy (or to another viral infection). This association can sometimes probably also be due to an infectious disease of the early childhood. Heredity seems to be a rather exceptional factor.

Concluding, we want to mention that in order to elucidate more precisely the importance of viral infections in the genesis of macular dystrophy associated with deafmutism, one has to examine systematically apparently normal and non deafmute children, born from mothers, who developed a viral infection during the first three months of pregnancy.

Summary

Most of the cases of Amalric's pigmentary macular dystrophy associated with deafmutism seem to be due to a rubella or to another viral infection of the mother during pregnancy. This association can sometimes probably also be due to an infectious disease of the early childhood. Heredity seems to be a rather exceptional factor.

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RIASSUNTO

La maggior parte dei casi di distrofia maculare pigmentaria del tipo Amalric associata a sordomutismo sembra dovuta a rubeola o ad altra infezione virale della madre durante la gravidanza. Questa associazione qualche volta può anche essere dovuta a malattie infettive contratte nella prima infanzia. L'influenza dell'eredità sembra eccezionale.

RESUMÉ

La plupart des cas de dystrophie maculaire du type Amalric associée à la surdi-mutité semblent dus à une rubéole ou à une autre infection virale de la mère pendant la grossesse. Cette association peut parfois être également due à une maladie infectieuse de la toute première enfance. L'hérédité paraît intervenir plutôt exceptionnellement.

ZUSAMMENFASSUNG

Es scheint, dass die meisten Fälle von Dystrophia macularis in Verbindung mit Taubstummheit durch Röteln oder andere Virusinfektion der Mutter während der Schwangerschaft bedingt sind.

Diese Verbindung lässt sich manchmal auch auf eine Infektionskrankheit im Laufe der frühen Kindheit zurückführen. Es scheint, dass die Mitwirkung eines Erbfaktors ziemlich aussergewöhnlich ist.