



## Miscellaneous Clinical Case Reports in Twins

L. Gedda, S. Berard-Magistretti, G. Brenci

*The Gregor Mendel Institute of Medical Genetics and Twin Research, Rome*

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**Abstract.** Three case reports are presented concerning 1) Albino MZ female twins, 2) Raynaud's syndrome in MZ female twins, and 3) Pseudohermaphroditism in MZ male twins.

**Key words:** Albinism, Raynaud's syndrome, Pseudohermaphroditism, Twins

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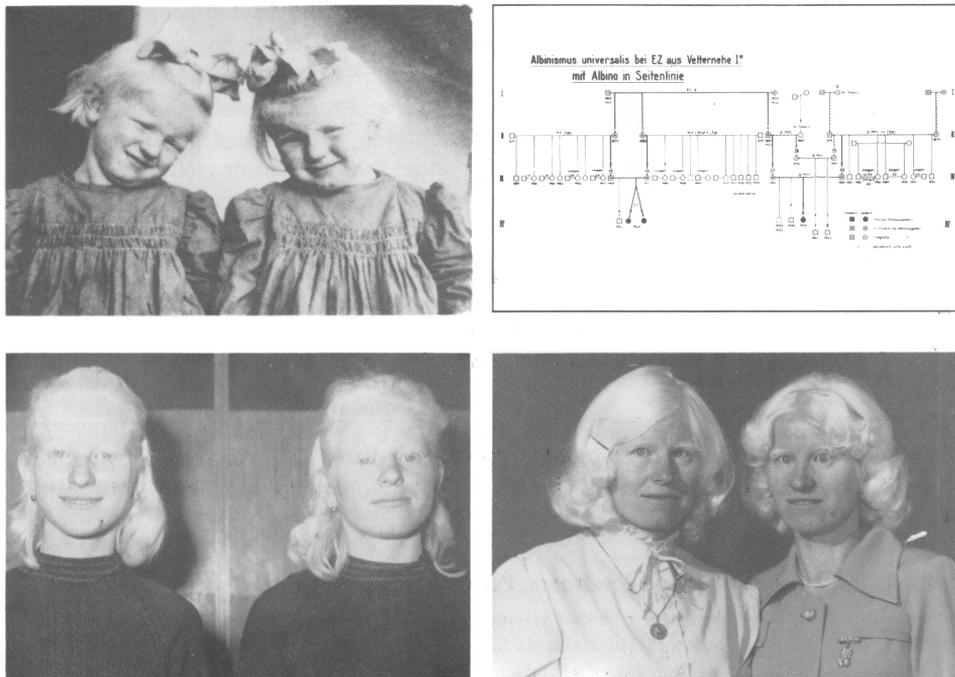
We shall present here three case reports of twins involving different conditions and that may prove of interest from various clinical perspectives as well as genetically.

### 1. ALBINO MONOZYGOTIC FEMALE TWINS

In 1953, Hanhart published in our journal a case of concordant total albinism in two MZ twin sisters of the Aosta Valley who were 7 years old at the time (Fig. 1). Their pedigree indicated that the parents were first-degree cousins and that a second-degree cousin of the twins was also affected by albinism (Fig. 2). As we later conducted a study on the twins of the 74 towns of the Aosta Valley, we established a regular contact with these albino MZ twin sisters, Anna and Maria, who were then 15 years old. Figures 3 and 4 show these twins at that time and as they are now, at the age of 37. Anna is the mother of two boys, and Maria of two girls.

Ophthalmological examinations have been made in the albino twins, in their mother, and in their respective husbands and children. Identical findings were obtained in the twins: absence of pigment from the fundus, which shows the choroidal and retinic vessels; horizontal nystagmus in small shocks; divergent strabismus, myopic astigmatism of 3 diopters in both eyes. Biomicroscopy showed absence of the posterior iridal pigment layer of ectodermal origin, while the anterior iridal layer of mesodermal origin shows absence of pigment. Moreover, in the iridochorneal corner, the anterior layer does not

stop at the basis of the iris but covers most of the muscle band. Interestingly, this latter finding, that is, the muscle band covered by the anterior layer, is also seen in the twins' mother, who is not otherwise affected by albinism, while no abnormality is found in the twins' husbands and children. It is the first time, to our knowledge, that this anomaly of the iridochoroidal corner is described in albino patients and in one of their relatives, the mother, who might be a heterozygotic carrier.

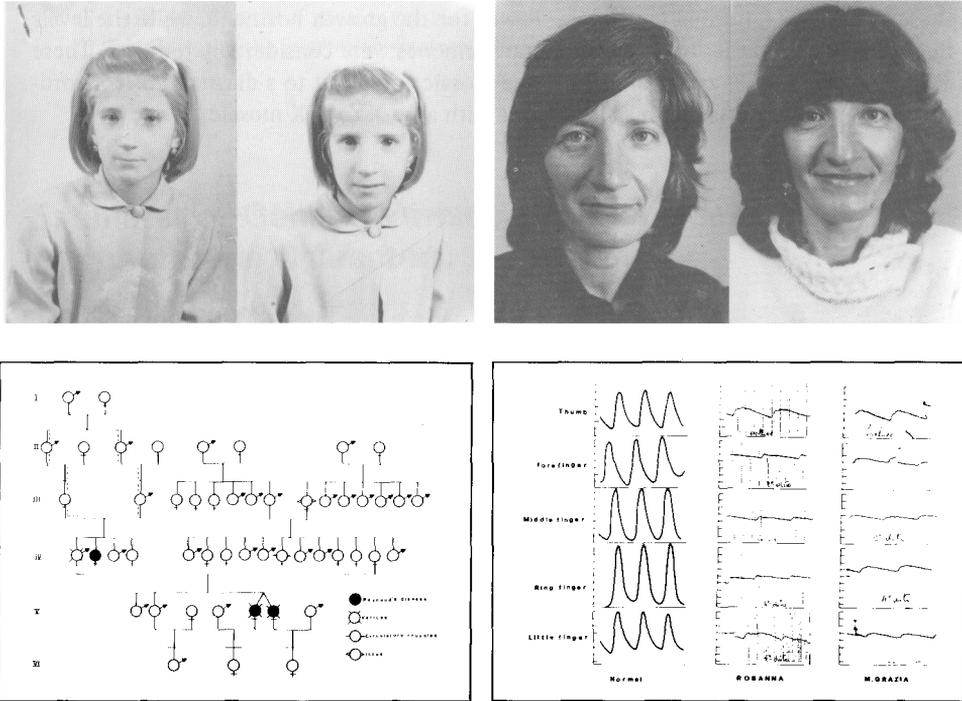


Figs. 1-4 - Concordant albinism in MZ twins (see text).

## 2. CONCORDANT RAYNAUD'S SYNDROME IN MONOZYGOTIC FEMALE TWINS

Our second case report concerns two MZ twins sisters concordant for Raynaud's syndrome. Rosanna and Maria Grazia were 11 years old (Fig. 5) when we first met them and are now 37 (Fig. 6). Both twins are married and each has a daughter. They live apart, do not get along and have financial contrasts. Rosanna is a housewife and lives in a low area, while Maria Grazia is a dish-washer in a hotel at about 300 meters of altitude. The symptoms of Raynaud's syndrome appeared in both twins around the age of 30. Their pedigree (Fig. 7) indicates the presence of the syndrome in the two twins, who are the last-born in the fifth generation, as well as in a paternal aunt – born, as the twins' father, of a consanguineous marriage between first-degree cousins. Other symptoms of the condition, such as varicose veins, circulatory troubles and ictus, are present in other relatives. Acroischemia of the left arm is present in both twins. Circulation in the fingers is illustrated by plethysmographic analysis (Fig. 8). Acroischemia is also present in the legs, where varicose veins are also evident in both twins. Finally, the X-ray analysis indicates laxity of carpal and radioulnar ligaments.

To our knowledge, family studies of Raynaud's syndrome are restricted to the two families described by Lewis and Pickering in 1933, and the only twin case report, described by Ullrich in 1926, refers to a discordant DZ pair. That is why we have considered it worthwhile to report on this case of concordant Raynaud's syndrome in MZ twins with one more case in family members.



Figs. 5-8 - Concordant Raynaud's Syndrome in MZ twins (see text)

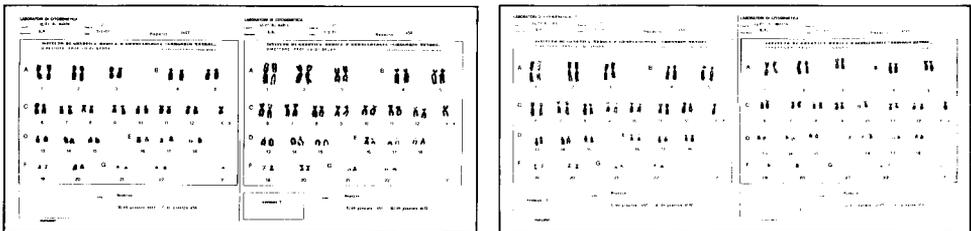
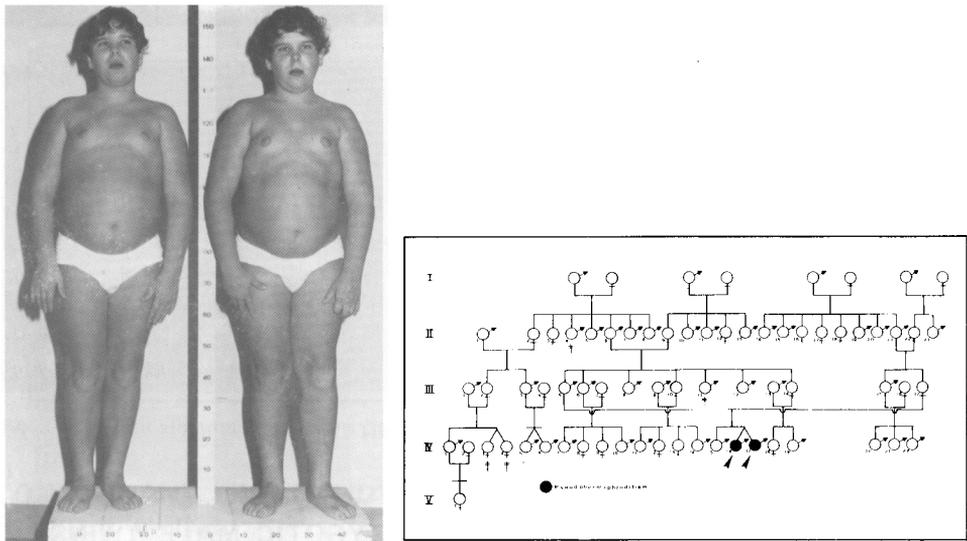
### 3. PSEUDOHERMAPHRODITISM IN MONOZYGOTIC MALE TWINS

Our third and final case report concerns two MZ twin boys concordant for pseudohermaphroditism. Castore and Polluce (Fig. 9) were referred to the Mendel Institute in 1981 with a diagnosis of "cerebroathic oligophrenia". They were 14 years old, respectively 159 and 157 cm of height. Both had a normal EEG. However, they had not reached puberty and presented atrophic male genitalia. The result of a previous chromosomal analysis, restricted to Polluce, had indicated a "normal male karyotype with no evidence of structural aberrations".

We started from the study of the pedigree (Fig. 10) and found the twins' parents to be second-degree cousins. One more pair of identical twins was present in the paternal side. The twins were born two years after a normal boy and when their father was 33 years old and their mother 22. Neuropsychiatric examination indicated psychomotor disturbances with a considerable delay in language development and slow learning processes. (They attend a special school, corresponding to a fifth grade in the regular primary school).

Our own chromosomal analysis indicated the presence of a mosaic in both twins, with a 45,X karyotype in 15% of Castore's cells (Fig. 11) and 20% of Pulluce's cells (Fig. 12). Similar findings characterize about 5% of cases of Turner's syndrome and about 50% of cases of pseudohermaphroditism. Hormone studies were carried out in the laboratories of the Endocrinology Chair of the University of Rome Medical School (Prof. Andreani). Normal levels were found for the thyroid and pancreatic hormones. Of the adenohipophysial hormones, normal levels were found for the growth hormone, while the levels of the luteinizing and the follicle-stimulating hormones were considerably reduced. These findings lead to exclude a Turner's syndrome mosaic and point to a diagnosis of concordant pseudohermaphroditism in male MZ twins with a 46,XY;45,X mosaic.

Correspondence: Prof. Luigi Gedda, Istituto Mendel, Piazza Galeno 5, 00161 Rome, Italy.



Figs. 9-12 - Concordant pseudohermaphroditism in MZ twins (see text)