THE WAARDENBURG SYNDROME

REPORT OF AN ABORTIVE CASE

F. P. Scott, M.Med. (Derm.), and J. A. van Beukering, M.D.

The National Hospital and the Postgraduate Steering Committee, Bloemfontein

Waardenburg,[1,2] after collecting material for many years in various deaf-and-dumb institutions in Holland, described a new syndrome in 1951. He called it 'a new syndrome combining developmental anomalies of the eyelids, eyebrows and nose-root, with pigmentary defects of the iris and head hair and with congenital deafness'.

The complete syndrome shows the following signs: (1) Lateral displacement of the medial acanthi, (2) prominent broad root of the nose, (3) approximation of eyebrow growth, (4) white forelock, (5) heterochromia iridium, and (6) deaf-mutism or incomplete congenital deafness.

Of the above abnormalities, the displacement of the eye-

lids seems to occur most frequently. The disease is inherited as an autosomal dominant with no significant sex differences. Mutation is responsible for sporadic cases, and the first mutant might show deafness or any combination of the various abnormalities. Waardenburg found 29 patients with this syndrome in Holland.

Congenital deafness occurs in association with other anomalies. Fraser *et al.*[3] described a clear genetic association between congenital deafness and goitre in 18 families, with 26 cases of goitre and deafness. They could demonstrate that this syndrome differs genetically from the Waardenburg syndrome. Fisch[4] also described a separate syndrome with early greying of the hair, heterochromia and deafness, which differs from the type of deafness in the Waardenburg syndrome. Hearing is affected over the whole tonal scale, while in the Waardenburg syndrome (where deafness is partial) it is affected mainly in the lower and middle ranges.

Ballantyne[5] also described a familial deafness with blonde hair and blue eyes, where there is a progressive loss of hearing affecting firstly the high tones.

CASE REPORT

The following case observed by us appears to be a sporadic example of an abortive Waardenburg syndrome; it concerns a Bantu male child of 11 years who, according to the parents, was totally deaf from birth. The boy's clear blue eyes are very unusual in a Bantu. He is from pure Bantu stock and definitely had no White ancestors. His features are also pure Bantu. The white guardian of the patient could give us a fairly accurate history of this child, who was practically brought up in her home. In spite of his deafness his intelligence was remarkable. Even while in hospital he was able to understand practically everything that was said to him, through lip-reading. Examination revealed a total deafness. (We wish to thank Dr. D. J. Franck for the otolaryngological examination.) There was no lateral displacement of the acanthi. His face showed a distinct prognatia; the nosetip was fairly sharp, but his alae nasi were typical Bantu. There were no other abnormalities such as sinusitis or nasal obstruction.

There was no white forelock, but at the first examination we found a single white hair in this area. The hair on the sides of the head showed a slight red discoloration and looked like the hair of children suffering from kwashiorkor. This child was definitely not underfed and, according to his guardian, he enjoyed the same privileges as her own children from birth. According to Waardenburg[6] the red hair discoloration might have some association with this syndrome because depigmentation or other colour anomalies usually seen on the forehead might also occur on other parts of the scalp. Further systemic examination revealed no congenital or other abnormalities.

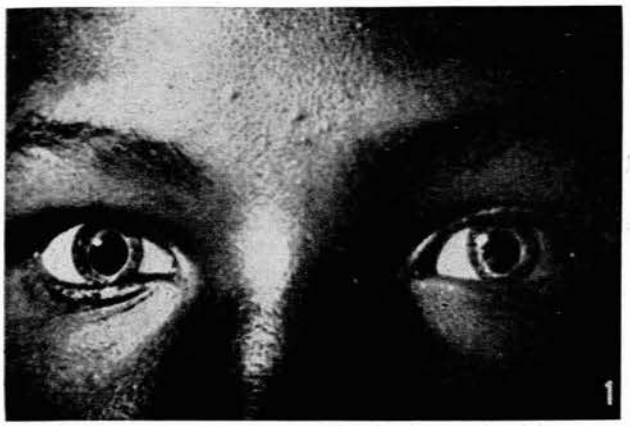

Fig. 1. Blue eyes in a Bantu child showing pseudo-coloboma.

We wish to thank Dr. G. L. Trichard for the ophthalmological examination. He reported as follows: 'A blue-eyed Bantu child with slight pseudo-coloboma bilateral at 6 o'clock. Iris architecture anti-clockwise spiral. Both irides show structural degeneration and pigment granules in the anterior layers. The fundi revealed a pigment migration not necessarily in any definite form. It does not follow the vascular tree. Both maculae are underdeveloped' (Fig. 1).

On examination the mother and other members of the family showed no signs of the syndrome. We were assured that the father was normal in all respects.

DISCUSSION

Waardenburg analysed the various families in his series genetically and could show that the inheritance is dominant. The genesis of the individual symptoms is not yet clear. The lateral displacement of the acanthi is nearly always present. It is absent in our case, but the remarkable association of blue eyes in a Bantu, deaf-mutism and red discoloration of the hair, makes it likely that this is an abortive case of the Waardenburg syndrome.

Fisch also mentioned 8 cases without the eyelid abnormality. He examined the temporal bone and whole auditory pathway in a female child of 3½ years suffering from this syndrome. Apart from an inflammatory reaction he found the only abnormality to be a complete absence of the organ of Corti; this has also been seen in cats with heterochromia of the iris, and Waardenburg discussed the family tree of a bullterrier family suffering from this syndrome.

According to Fisch the white forelock is situated at the site of the third median eye of early vertebrate development. Both the auditory apparatus and the pigment cells develop from the neural crest and form part of the protective function of the organism. A white forelock is not always associated with this syndrome, however. We have seen a male patient belonging to a family in which the white forelock had been present for many generations without deafness or any of the other signs of the Waardenburg syndrome.

One of us (J.A.v.B.), while practising in Ghana, saw heterochromia iridis in 2 Negro sisters, and red hair in a pure Negro boy. It was not possible to examine other members of these families. No other congenital abnormalities were noted in these patients.

The Waardenburg syndrome must be borne in mind when blue eyes are noted in pure Negroes.

REFERENCES

1. Waardenburg, P. J. (1951): Amer. J. Hum. Genet., **3,** 195.
2. *Idem* (1957): Acta ophthal. (Kbh.), **35,** 311.
3. Fraser, G. R., Morgan, M. E. and Trottes, W. R. (1960): Quart. J. Med., **29,** 279.
4. Fisch, L. (1959): J. Laryng., **73,** 355.
5. Ballantyne, J. C. (1960): *Deafness.* London: Churchill.
6. Waardenburg, P. J.: Personal communication.