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A PIEBALD FAMILY.

By E. A. COCKAYNE, M.D., M.R.C.P.

In spite of the great interest, which they have always excited, well authenticated examples of piebalds in the dark races have been found to be rare. In the white races they are much less conspicuous, in part owing to the presence of clothing and in part owing to the lack of contrast between the pigmented and unpigmented skin, but the likelihood of their coming under the notice of a skilled observer is much greater. The scarcity of records shows that piebalds in the white races also must be very uncommon. Last year I met with a case in a baby, and found that the child belonged to a family, many of whose members showed a similar defect of pigmentation. The family, belonging to a farming stock, originally came from the neighbourhood of Bury St Edmunds in Suffolk, and the anomaly is known to have descended directly through six generations. The oldest member, with whom I have talked, is fairly certain that it was present in one generation at least before this.

Of the first two generations in the pedigree (see Plate XI), I could obtain no definite information except the statement as to the existence of the piebaldism in I. 1 and II. 1, but of the third, III. 2 is said to have had a frontal blaze of white hair and white skin on the neck and forearms, which was very conspicuous owing to its marked contrast with the neighbouring weather-stained normal skin.

III. 4 appears to have been the only member of the family who showed a marked dislike to the condition and always wore a wig to hide the frontal blaze. III 2, whose family name was C——*, had fifteen children. The first, IV. 2, a male, with dark hair, married twice, and had eight normal children, five by the first wife and three by the second. The second child, IV. 5, was a piebald, with a large frontal blaze, white skin on the front of the neck and arms, and blue eyes. He transmitted the condition to all his three children. V. 3, the eldest boy, aged 22 and unmarried, possesses dark hair, with a V-shaped frontal blaze of white or cream coloured hair, the apex of the V commencing near the coronal suture and spreading out to a width of $3\frac{1}{2}$ inches, as it reaches the forehead. The eyebrows and some of the eyelashes are white. The next boy, V. 4, is aged 18. He has light hair and a very large blaze of unpigmented hair, which covers the whole of the top of the head. His eyebrows and eyelashes are white, and the eyes are blue. Both boys have white patches on the front of the neck and on the arms (see Plate XII).

* Names preserved in the confidential register of the Galton Laboratory.

Next in the fourth generation were twins, IV. 6 and 7, both piebalds. They were evidently not uniovular, because one had dark hair, and one light, and the white blazes were dissimilar in extent, but it is uncertain which had the larger. Both died at an early age.

The next, IV. 8, a girl, was normal with dark hair and eyes and remained unmarried. Next came a woman, IV. 10, who was a piebald with a large frontal blaze, white eyebrows and eyelashes, and white skin on the front of the neck and forearms. *The right eye was blue, and the left brown* (see Plate XIII (B)). Her child, aged 13, is quite normal with light hair and dark eyes.

The next child, IV. 12, Mrs W——, has a large frontal blaze and dark brown irides. There is a large irregular patch of white skin extending from just below the chin to the heads of the clavicles, and round it the skin appears to be more deeply pigmented than the rest of the skin of the neck. There are a few small islands of pigmented skin near the edge of the unpigmented area. The skin of the anterior aspect of the forearms is unpigmented from the elbows to the wrists, and here also, there are some small islands of pigmented skin in marked contrast to the unpigmented area, in which they lie (see Plate XIV).

The first two children of this individual were daughters, V. 8 and V. 9, both piebald, the third a normal son, V. 10, and then three more piebald daughters, V. 11—13. The first of the daughters, Mrs G——, V. 8 (see Plate XIV), is very fair with a very large frontal blaze covering the whole of the top of the head, and her eyebrows and eyelashes are white. Her normal hair has pale creamy diffused pigment and, according to the individual hair, some to a decided number of granules*. The hair of the blaze has no diffused pigment and no granules. The irides are light brown, but the outer segments on both sides are paler and greenish in colour. The skin of the forehead and base of the nose is very pale in colour. She has a large white patch on the skin of the front of the neck, beginning just below the chin and widening out so as to embrace that over the inner ends of both clavicles. As in her mother there appears to be some concentration of pigment round this white area, and there are small isolated areas of pigmented skin near its edge. She has unpigmented skin on the anterior aspect of both forearms.

Of her two children the first, VI. 1, a boy aged 3, is normal, the second, VI. 2, a boy aged 1½, is a piebald (see Plate XIV). This child, VI. 2, was nine months old when first seen. He had a very large frontal blaze, resembling that of his mother and covering all the top of the head, the eyebrows and eyelashes were white with the exception of some of the outer hairs. Hair, pale cream in colour, said to be from the light area, has pale creamy diffused pigment and some granules (β), the granules being very small. It was obvious, even at this age, that heterochromia iridis was present. The right iris was pale except for a sector of dark grey occupying the upper and outer quadrant, the left iris was entirely dark grey. No difference in colour of the skin of the neck or forearm could be made out.

* β to γ on the Galton Laboratory scale of granular pigmentation.

When the baby was seen after the summer of 1913, the grey portions of the irides were becoming brown, the pale portion was still light blue. The face and arms were sunburnt, and it was noticed that the forehead was paler than the rest of the face. There was a pale area on the front of the neck, and the whole anterior surfaces of the forearms were white, the edges being very irregular in contour. There was also a white streak running obliquely right across the posterior or extensor aspect of the left forearm, and this offered a marked contrast with the rest of the surface, which was very brown. When the sunburn had died away the difference between the pigmented and unpigmented skin could no longer be made out.

IV. 12's second daughter, V. 9, aged 23, has only a small cream coloured frontal blaze, and the rest of her hair is light brown (see Plates XV and XVI). The eyebrows are composed of an even mixture of brown and white hairs, and the eyelashes are similar, with brown and white hairs alternating. The irides are grey and uniformly pigmented. There is a large irregular area of white skin at the base of the neck.

The whole of the anterior aspect of the right forearm is unpigmented, and there are similar small areas scattered over the posterior aspect (see Plate XVII). The left forearm is white only on the anterior aspect.

The next girl, V. 11, is aged 9. She has a very small frontal blaze, but the skin of the forehead is pale (see Plate XV). The eyebrows show a division into two parts, on the inner halves grow white hairs only, and on the outer brown hairs. The eyelashes on the contrary consist of alternate brown and white hairs. The irides are grey and uniformly coloured (see Plate XVIII). There is only a small white area in the middle of the front of the neck, but there are well differentiated white areas on the anterior aspects of both forearms (see Plate XIII (A)), and on the inner aspects of both upper arms. Her hair was examined and the first sample showed very pale diffused pigment and some granules (β). Two more samples were then examined, one from the blaze and one from the neighbouring part of the scalp. The first showed no diffused pigment and no granules, the second showed the majority of hairs with yellow-brown diffused pigment and a decided number to plenty of small granules (γ — δ), but a few had no diffused pigment and no granules.

The next piebald child, V. 12, died young. She had a frontal blaze and blue irides. Some of her hair showed very pale diffused pigment, and some granules (β).

The next child, V. 13, also died young. She was a piebald nearer to the classical type than any of the others. She had a large frontal blaze, white skin on the forehead, and large areas of white skin on the front of the neck and chest, and in addition a very extensive area on the abdomen.

Of the fourth generation the next child, IV. 13, was a male with dark hair and eyes, who had 5 normal children; the next, IV. 15, had fair hair and died young. Twins, IV. 16 and 17, came next and died in infancy*. They were heterogeneous,

* The tendency to twin in this family is worth noting.

a dark-haired boy and a light-haired girl. A girl, IV. 19, was born next and she had twin sons, V. 15 and 16, who were also normal. The last three children, IV. 20—22, a girl, a boy, and one whose sex I am unable to ascertain, were all normally pigmented and all died at a very early age.

The pedigree confirms the strongly hereditary nature of piebaldism, and in this as in other published cases the character can affect either sex, but has only been transmitted by those affected. Unless we are to assume that in the case of such a rare anomaly as piebaldism, I. 2, II. 2 or III. 1, were really unnoticed piebalds, then III. 2 could only be heterozygous, or since piebaldism is dominant a (*DR*). We must take IV. 4, IV. 9, IV. 11 and V. 7 for pure recessives (*RR*). Thus the number of piebalds in the five sibships of generations IV, V and VI should be one quarter, i.e. $\frac{1}{4}(15 + 3 + 1 + 6 + 2) = 7$ nearly. We have actually 14 out of 27, thus piebaldism does not seem to act numerically as a pure dominant.

The areas of unpigmented skin are less than in the classical piebalds, but it is probable that in some, at least, they are larger and more numerous than I have stated. On the covered parts of the body and legs, which I was unable to examine except in the baby, they would not be very noticeable. It was not until I had noticed the white skin on the neck and arms of one of them that I was told anything about the existence of similar patches on the others. If true, it is remarkable that none have had white patches on the legs.

With regard to the local distribution of the pigment, there appears to be an excess at the edges of some of the unpigmented areas, as has been noted in other cases. In the case of other pale areas, the demarcation between them and the normal skin is very slight, and is probably due to the fact that they are not wholly unpigmented. This remark applies especially to the forehead, which in some of them looks paler than natural, but not wholly devoid of pigment.

In some the eyelashes are alternately white and brown, and in others the eyebrows are similar, and in one at least hairs growing on the scalp near the blaze are in some instances entirely without either diffused or granular pigment. This suggests that the skin beneath may show a deficient and irregular distribution of pigment.

The most interesting feature is the occurrence in three members of the family of well-marked heterochromia iridis, a character which has been met with in members of a piebald family, but always independently of their piebaldism, never, as in this case, in true association with it. It proves conclusively that these cases are not congenital leucoderma.

There seems to be no association of piebaldism and general lack of pigmentation of hair and irides. Affected and unaffected members have been both fair and dark, but the fairest piebalds seem to have the most extensive frontal blazes.

In the cases photographed the individuals were blonds and there has been great difficulty in getting a good photographic contrast of differences of pigmentation very noticeable in the living subject.

Pedigree of Piebald Family.

V. 3 and V. 4 as children showing their marked V-shaped frontal blazes.

B

IV. 10. The blaze may be seen with difficulty under a lens, but the heterochromia iridis is easily distinguishable.

A

V. 11. Showing a well differentiated leucotic area on the anterior aspect of the left arm.

Piebaldism in three generations, V. 8, mother, IV. 12, grandmother, and VI. 2, grandson. The white frontal hair is visible in all three and the darker section of the right eye of VI. 2 with the use of a lens.

V. 11 and V. 9. Two sisters with white frontal blazes.

V. 9. Showing white forelock or blaze.

Right forearm of V. 9 showing white patches on posterior aspect. The photograph is untouched and it is difficult to bring out by photography the grades of pigmentation when the arm is untanned by the sun, although they are quite clear on actual inspection.

Large photograph of V. 11 to show paleness of forehead and white hairs on inner half of eyebrows.