



## Case Report

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# Piebaldism: about an Observation and Review of the Literature

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Email: drtshilombo@gmail.com**Abstract**

The authors describe a case of piebaldism in a child of 1 year and a half and make a brief review of the literature. Piebaldism is a rare and benign condition. His diagnosis is clinical, after elimination of other differential diagnoses.

**Keywords:** Piebaldism; Depigmentation; Skin; Hair; Hypomelanosis**Introduction**

The presence of localized hypomelanosis of the skin and hair in a newborn should lead the clinician to evoke a rare genetic disease such as piebaldism. We report here a case of piebaldism observed at the University Clinics of Kinshasa and address, through a review of the literature, its epidemiological, clinical and therapeutic aspects.

**Observation**

**Figure 1:** White lock on the front and hypochromic macules on the lower limbs in our patient.

IM, 1½ years old, is the result of a non-consanguineous marriage. She is born to term and has a psychomotor development normal for her age. This child was followed since birth for multiple hypopigmented lesions. The heteroanamnesis reveals the presence of the same abnormalities in his great-

grandmother, without association with other pathologies. At the clinical examination, it was a child in good condition with hypopigmented tablecloths of interest to the lower limbs dotted with normally pigmented skin. In addition, the child also had hypopigmented triangular hair involvement in the mid-front scalp. ENT, ophthalmic and cardiovascular examinations were normal. On all of these clinical data, the diagnosis of piebaldism was retained.

Protection advice for hypopigmented areas of the skin as well as the application of a cream with sunscreen have been prescribed to avoid complications (Figure 1).

**Discussion**

Piebaldism has been described for a long time throughout history with early writings by Egyptians, Greeks and Romans [1,2]. It is a pigmentation disorder of genetic origin with autosomal dominant inheritance [3]. It is a mild, rare disease whose incidence is estimated at less than 1/20000 live births

[4,5]. This pigment anomaly results from a mutation in the proto-oncogene KIT at chromosome 4, or in the gene SLUG at chromosome 8; these mutations are responsible for a lack of migration and differentiation of melanoblasts during embryogenesis [6-8].

Clinically, this syndrome is manifested by the presence, at birth, depigmented spots that may interest the face, chest, abdomen, extremities. These spots are stable and persistent with distribution symmetry [9]. A lock of white hair, often triangular in shape, is present on the front of the scalp, sometimes with the underlying forehead [10]. In 80 to 90% of cases, this frontal white lock may be the only clinical manifestation [2,11]. Eyelashes

and eyebrows can also be achieved. Typically, additional hyperpigmented macules can develop in the white plaques [9].

Diagnosis of piebaldism is clinical after elimination of other differential diagnoses. The main differential diagnoses are albinism, Waardenburg syndrome, tuberous sclerosis, vitiligo, Vogt-Koyanagi-Harada syndrome, Alezzandrini syndrome, alopecia areata and sarcoidosis [4]. Rare cases of association of piebaldism with other diseases, such as Hirschprung's disease, neurofibromatosis type 1, Blackfan-Diamond anemia, glycogenosis type 1a have been described in the literature [4-5,12].

From a therapeutic point of view, areas of skin depigmented in piebaldism are generally insensitive to drug treatments and phototherapy [4], but it is important to protect them from sunstrokes by using protective creams to avoid a transformation. malignant [11]. Transplantation of the skin with the aid of the technique of mini-grafting, effective in 80% of cases, would currently be the best alternative [5].

### Conclusion

Piebaldism is a mild illness but can have psycho-social repercussions. Therapeutic research is underway, including genetic therapy to improve the quality of life of patients.

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