Piebaldism: A rare presentation in Nepalese context

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Abstract

Piebaldism is a rare autosomal dominant disorder characterised by a congenital white forelock and multiple symmetrical hypopigmented or depigmented macules. We present a case of an 18 year old male with a typical clinical presentation, followed by a concise review of the literature discussing the genetics, clinical features, diagnosis, and management of the condition. This is the first case of Piebaldism to be reported from Nepal with a good cosmetic surgery outcome.

Key words: Autosomal dominant, Hypopigmentation, Piebaldism

INTRODUCTION

Piebaldism is an autosomal dominant genetic disorder of pigmentation characterised by congenital patches of white skin and hair that lack melanocytes[1]. The hypopigmented areas usually present as white fore lock and leukoderma on the frontal scalp, forehead, ventral trunk and extremities. The white hair and patches of such patients are completely formed at birth and do not usually expand thereafter. It results due to the absence of melanocytes in affected skin and hair follicles due to mutations of the *KIT* proto-oncogene, which encodes the cell-surface receptor transmembrane tyrosine kinase for an embryonic growth factor[2].

CASE REPORT

An eighteen year old gentleman from eastern Nepal presented at Kathmandu Medical College Teaching Hospital (KMCTH), on November, 2013 with chief complaints of hypopigmented area on the forehead, which was present since birth. The patient had come for excision of the hypopigmented area primarily for cosmetic purpose. He was an average built teenager, was in usual state of health and had no other systemic complaints. His mother, maternal uncle, and some of his cousins also suffered from similar conditions. On examination, his general condition was good and his vitals were within normal limits. His systemic examination revealed no abnormality. He had areas of hypopigmentation over the forehead, bilateral lower limbs, chest and abdomen. On local examination, there was an area of hypopigmentation on his forehead, triangular in shape, present in the midline around 4×3 cm, and a forelock was present in continuity with the hypopigmented area (Figure 1). The macules were present since birth and were non-expanding in nature. The patient had similar areas of hypopigmentation with patchy islands of hyperpigmented macules within the area of hypopigmentation on the chest, abdomen and bilateral lower limbs of variable shapes and sizes (Figure 2, 3). He had a provisional diagnosis of piebaldism. The patient was then planned for the excision of the hypopigmented area of the forehead over two sittings. The following day, the patient was operated. Around 60% of the hypopigmented lesion was removed in first sitting and the remaining portion was planned to be removed in next operation six months later. Inner suturing was done with 4-0 monosyn while skin was sutured with 5-0 prolene (Figure 4). The excised tissue was sent for histopathological examination. On microscopic examination, the sections showed skin and underlying subcutis. Epidermis showed significant reduction in melanocytes throughout its length with few patchy deposits of melanin pigment. The dermis showed skin adnexal structures and was unremarkable. The histopathological features were compatible with clinical diagnosis of piebaldism (patterned leukoderma) (Figure 5, 6). When he visited the hospital five months back, (Figure 7) he was very satisfied with the surgical outcome and though serial excision was planned for the patient in the subsequent visit it was not deemed necessary.

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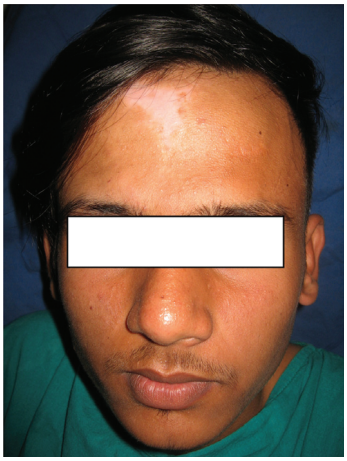
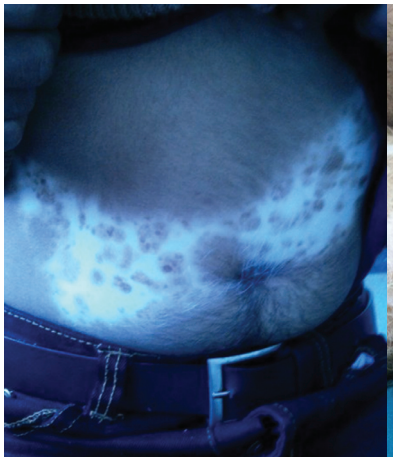


Figure 1: Area of hypopigmentation on forehead and a white forelock present in continuity with the hypopigmented area.



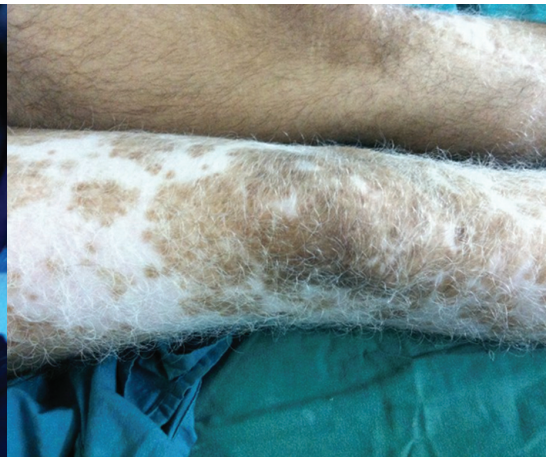


Figure 2, 3: Areas of hypopigmentation with patchy islands of hyperpigmented macules within the area of hypopigmentation on the abdomen and bilateral lower limbs of variable shapes and sizes.

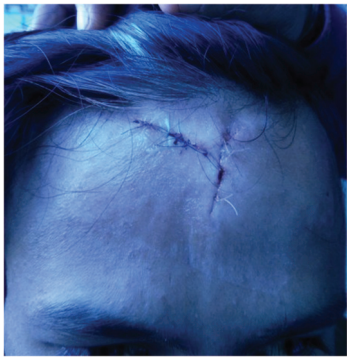
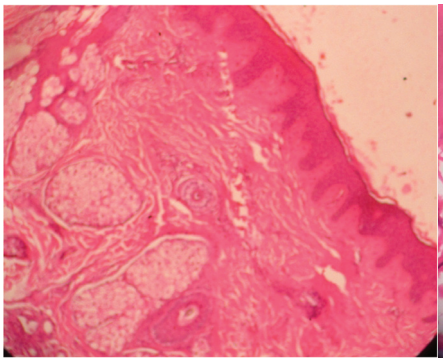


Figure 4: The areas of hypopigmentation removed and sutured.



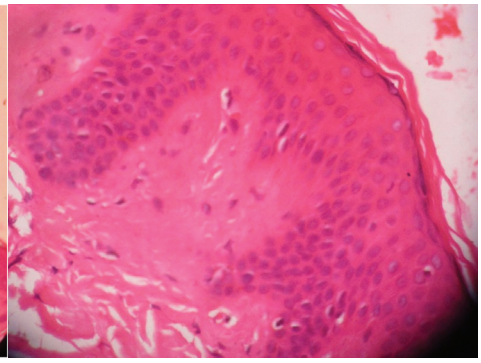


Figure 5, 6: Hisotpathological section (10×, 40×) showing patterned leukoderma.

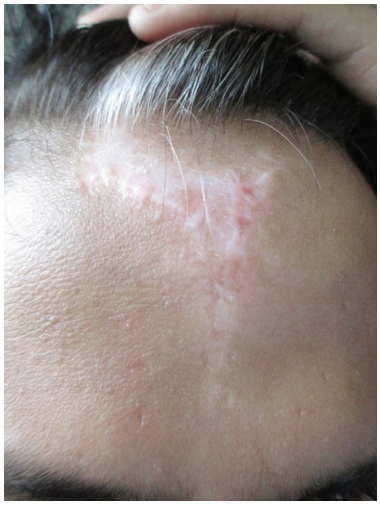


Figure 7: The post operation scar five months later.

DISCUSSION

Piebaldism is an autosomal dominant genetically inherited pigment anomaly characterised by congenital white skin (leukoderma) and white hair (poliosis) on the frontal scalp, forehead, ventral trunk and extremities. It occurs due to mutations of the KIT proto-oncogene, which encodes the cell-surface receptor transmembrane tyrosine kinase for an embryonic growth factor[3]. The severity of phenotypic expression in piebaldism correlates with the site of mutation within the KIT gene[4]. The most severe mutations seem to be dominant negative missense mutations of the intracellular tyrosine kinase domain, whereas mild piebaldism appears related to mutations occurring in the amino terminal extracellular ligand-binding domain with resultant haplo insufficiency[5]. Piebaldism may present alone or sometimes it could be one of the cutaneous signs of Waardenburg syndrome[6]. A number of syndromes associate piebald-like hypopigmentation of the skin and

hair with other anomalies, but are not associated with anomalies of the KIT gene. In Waardenburg syndrome, along with the dermatological manifestations of piebaldism, heterochromia of the bilateral iris, lateral displacement of inner canthi of eye, and sensorineural deafness may also be present[7,8]. Although piebaldism may visually appear to be partial albinism, it is a fundamentally different condition. The vision problems associated with albinism are not usually present as eye pigmentation is normal in Piebaldism[9]. Ziprkowski and Margolis in 1962 described an X-linked recessive disorder characterized by hypomelanosis, deafness and mutism in a Jewish Israeli family of Sephardic origin[10]. It has now been included in the albinism-deafness syndrome (ADFN) and the gene has been localized to Xq24-q26 but not identified[11]. Woolf first reported piebaldism in association with congenital deafness, in 1965 in two Hopi Indian brothers in Arizona[12]. The Tietz syndrome was first described as a congenital generalized depigmentation and profound congenital sensorineural deafness, transmitted as an autosomal dominant trait with full penetrance, and attributed to a mutation in the micropthalmia-associated transcription factor (MITF) gene in descendants of the same family[13].

There are various treatment options available for piebaldism. Camouflaging could be considered as one of the main treatment options for patients with piebaldism as the areas of hypopigmentation are non-expanding in nature and are unresponsive to light or medical treatment[14]. Besides, other treatment options include surgical procedures like excision of the affected area of the skin or dermabrasion followed by skin grafting. Depigmented areas may be treated with thin split-thickness grafts and minigrafting or with in vitro cultured epidermis and suction epidermal grafting with additional minigrafting[15]. Recently, transplant of autologous melanocytes obtained through the culture of melanocytes or of melanocytes and keratinocytes has been described as a safe and effective treatment for patients with piebaldism[16,17]. This induced scar less repigmentation using a small donor site. The area then could be further treated with phototherapy for uniform repigmentation. However, in a resource poor setting like ours, camouflaging could be a better option than surgery as it is an easy yet highly effective method. Further management include genetic counselling of the patient to improve the quality of life for affected individuals.

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