



A Case of a 4-Year-Old Carbon Baby: Acquired Universal Melanosis and Literature Review

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Abstract

Disorders of color variation can be localized or generalized, congenital or acquired, acute or chronic. Among the various causes of generalized skin darkening encountered in the pediatric age group, acquired universal melanosis aka Carbon Baby Syndrome is an uncommon hypermelanotic condition, with a histologic appearance of extensive epidermal melanization. The authors report one such case of a 4-year old boy with progressive darkening of skin starting at the age of 1 year and involving the entire body surface in the absence of any other systemic alterations. The disease prevalence, etiology, mechanism and prognosis are still unknown.

Keywords: Carbon baby, Progressive pigmentation, Universal acquired melanosis

Introduction

Skin color is mainly dependent on melanin pigment that is produced inside melanocytes and is then transferred to the surrounding keratinocytes^[1]. Different skin tones are produced due to difference in the amount and forms (pheomelanin and eumelanin) of melanin produced by human melanocytes^[2]. The rate limiting enzyme in melanogenesis is tyrosinase, being the determinant of color, deciding varying shades of the iris, hair and skin^[3].

Acquired universal melanosis is a chronic progressive disorder of diffuse muco-cutaneous hyperpigmentation without any other systemic symptoms. There is increased melanin production and deposition in the basal and supra-basal epidermal layers by normal appearing (no alteration in shape or size) and normal number of melanocytes.

Case Presentation

A 4-year-old Indian boy born to consanguineous parents presented to the pediatric outpatient clinic with a history of progressive darkening of skin. It started appearing on the cheeks and feet at the age of 1 year, which then progressed to slowly involve the entire

body surface area. The prenatal, natal and postnatal periods were uneventful. His mother consumed prenatal vitamins and had no history of illness or drug intake. The child achieved his developmental milestones as expected for his age and has received all age-appropriate vaccinations. There was no history of fever, drug intake prior to appearance of discoloration, urine discoloration, colored body secretions, muscle weakness, nausea/vomiting, abdominal pain, itching, photosensitivity, prior skin infection, any behavioral problems, repeated blood transfusions. There was no similar history in the elder sibling or any other first/second degree relatives.

On examination, there was generalized hyperpigmentation of the body including the palms and soles but sparing the mucous membranes [Figure 1,2,3,4]. There was no difference in the intensity of hyperpigmentation between sun-exposed and non-exposed areas. There were a few areas of normal skin pigmentation on the face. The scalp hairs were normally pigmented and had no shaft abnormalities. The patient had normal skin texture with no skin lesions or nail abnormalities. On ophthalmologic examination, the fundus and retina appeared normal with no change in iris pigmentation. The vitals were within their normal limits as per age-appropriate cutoffs. Growth parameters- weight, height, BMI and upper limb: lower limb ratio was within their expected ranges. Examination of other organ systems was unremarkable.



Figure 1: Demonstrating the skin hyperpigmentation at the face, chest and abdomen.



Figure 2, 3 & 4: Demonstrating the cutaneous hyperpigmentation in AUM involving hands, feet and axilla.

Routine laboratory investigations were within normal limits. Other investigations including urine amino acids, serum iron studies, serum adrenocorticotropin hormone (13.70 pg/ml, Normal range 0-46 p/ml), very long chain fatty acid levels (VLCFA) revealed no abnormalities. An abdominal ultrasound was done which revealed no organomegaly. A punch biopsy of the hyperpigmented skin from the left leg showed epidermal atrophy with the presence of coarse melanin pigment in all layers of the epidermis. The dermis showed pigment incontinence with perivascular macrophages. Electron microscopy and chromosomal analysis could not be done because of lack of resources.

We treated our patient symptomatically with sunscreens to avoid UV radiation. The family was counselled regarding the condition from the limited reported data that could be gathered about it.

Discussion

There are numerous causes of generalized mucocutaneous hyperpigmentation which should be excluded before making a

diagnosis of AUM. Adrenoleukodystrophy causes progressive hyperpigmentation but spares palms and groin. Drugs (like clofazimine, anti-malarials) and heavy metals (like mercury, silver) can cause hyperpigmentation especially in children who have immature liver function, more body fat proportion and increased likelihood of accidental ingestion. Bronze baby disease is a condition due to abnormal photo isomer of bilirubin or porphyrin with dark grey-brown pigmentation of neonates with hepatic dysfunction undergoing phototherapy. Post-inflammatory hyperpigmentation occurs secondary to tinea infections or chronic bullous disease. Malnutrition like kwashiorkor, vitamin deficiencies and deposition disorders like haemochromatosis can also lead to generalized hyperpigmentation.

Familial progressive hyperpigmentation is described as patches of hyperpigmentation which are present from birth itself and grow in number and size progressively to involve the entire mucocutaneous surfaces. On microscopy, the melanin granules are more in number and larger in size than normal^[4]. Congenital diffuse melanosis is a disorder with hyperpigmentation developing soon

after birth and invading progressively to the trunk and limbs, with pigmentation being diffuse in the abdomen and reticulated in the neck and groin region. On electron microscopy, the melanosomes were not grouped within the keratinocytes but dispersed in the epidermal cell cytoplasm^[5].

Ruiz-Maldonado et al reported the first known case of acquired universal melanosis in 1978 and named it as “Carbon baby syndrome”. It was a Mexican-born Caucasian male who developed progressive blackening of skin, complete by the age of 4 years. Histology showed normal number of melanocytes with extensive epidermal and minimal dermal pigmentation. Electron microscopy revealed negroid pattern in the epidermal melanosomes^[6]. In 2008 and 2012, two cases of a 3-year-old girl and a 5-year-old girl were reported from India by Kaviarasan et al^[7] and Kumar et al^[8] respectively.

In 2012, Chakraborti et al^[9] for the first time reported this disease entity in two Indian siblings (brother and sister) from non-consanguineous parents, with the darkening starting at the age of 6 months. Findings of shortening and blunting of rete ridges with presence of stratum basale layer was observed on skin histology. In the same year, Shome et al^[10] reported the disease in two Indian siblings (5-year-old male child and 3-year-old female child) from consanguineous parents who were developing progressive skin darkening since the age of 5 months and 4 months respectively. Histology showed increased melanin in basal and suprabasal layers of epidermis and few macrophages in the dermis. In 2013, this condition was reported in a 3-year-old Japanese boy and a 4-year-old Iranian girl by Niiyama S et al^[11] and Toossi et al^[12] respectively. In 2014 Ghosh SK et al^[13], reported it in a young boy from India.

In conclusion, the exact etiology of AUM remains unknown. The following etiological hypothesis can be proposed for its occurrence. Sun exposure can be one of the environmental factors in the disease causation. The correlation has been supposed since greater number of cases have been reported from a tropical country like India, and some reported cases have described more pigmentation in sun-exposed parts as compared to non-sun exposed parts. Another possibility is some genetic mutation which may be involving recessive linked genes. This is based on the fact that disease occurrence is reported in siblings from both consanguineous and nonconsanguineous parents. Further studies are required to better understand the disease process, its pathophysiology, associated abnormalities and long-term prognosis.

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Conflicts of Interest

All authors declare no conflicts of interests.

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References

- [1] D'Mello SA, Finlay GJ, Baguley BC, Askarian-Amiri ME. Signaling Pathways in Melanogenesis. *Int J Mol Sci.* 2016; 17(7):1144. Published 2016 Jul 15. doi:10.3390/ijms17071144
- [2] Lin J.Y., Fisher D.E. Melanocyte biology and skin pigmentation. *Nature.* 2007; 445:843–850. doi: 10.1038/nature05660.
- [3] Videira I.F., Moura D.F., Magina S. Mechanisms regulating melanogenesis. *An. Bras. Dermatol.* 2013; 88:76–83. doi: 10.1590/S0365-05962013000100009.
- [4] Chernosky ME, Anderson DE, Chang JP, Shaw MW, Romsdahl MM. Familial Progressive Hyperpigmentation. *Arch Dermatol.* 1971; 103(6):581–598. doi:10.1001/archderm.1971.04000180007002
- [5] Kint A, Oomen C, Geerts ML, Breuillard F. Congenital diffuse melanosis. *Ann Dermatol Venerol* 1987;114:11-6
- [6] Ruiz-Maldonado R, Tamayo L, Fernandez-Diez. Universal acquired melanosis. The carbon baby. *Arch Dermatol* 1978; 114:775-8.
- [7] Kaviarasan PK, Prasad PV, Joe JM, et al. Universal acquired melanosis (Carbon baby). *Indian J Dermatol Venereol Leprol.* 2008; 74:38-40.
- [8] Kumar P, Ghosh K, Maiti A. Carbon baby syndrome (universal acquired melanosis). *Indian Pediatr.* 2011; 48:967-8.
- [9] Chakraborti S, Ghosh TN, Nag SS, et al. Universal acquired melanosis in siblings. *Indian J Pediatr.* 2012; 79:1094-6.
- [10] Shome K, Seth J, Samanta AB, et al. Carbon baby syndrome: two case reports. *J Pak. Assoc Dermatol* 2012; 22:59-62.
- [11] Niiyama S, Bando Y, Ishii M, Katsuoka K. Universal acquired melanosis: carbon baby. *Dermatol Online J.* 2013 Jul 14; 19(7):18961.
- [12] Parviz Toossi, Sarah Ershadi, Ehsan Abolhashmi. Acquired universal melanosis (carbon baby syndrome) in a 4-year old girl. *Iran J Dermatol.* 2013; 16:162-164.
- [13] Ghosh SK, Ghoshal L, Bhunia D, Ghoshal AM. Acquired universal melanosis (carbon baby syndrome). *Pediatr Dermatol.* 2014 Sep-Oct; 31(5):620-2. doi: 10.1111/pde.12077. Epub 2013 Jan 17.