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Birthmarks in Children

Identification and treatment of
birthmarks of medical significance

by Dr Lynn Chiam

Birthmarks are common and most pose no serious health risk. Birthmarks are not necessarily seen at birth and some manifest only within the first few weeks to months of life. Though most birthmarks are isolated skin conditions, they can uncommonly serve as an important diagnostic aid for other more significant disorders.

Congenital Melanocytic Nevi (CMN)

Approximately 1% of all newborns have a small CMN. Traditional classification of CMN in adulthood includes small (< 1.5cm), medium (1.5cm-1.9cm) and large (> 20cm). In a neonate, lesions measuring greater or equal to 9cm on the head or 6cm on the body can be considered large CMN.¹ Giant CMN typically involves significant portions of the skin, such as on the posterior torso, an entire extremity, or scalp. Giant CMN occurs in 1 in 500,000 compared to 1 in 20,000 for a large CMN.²

Large and giant CMN are associated with significant cosmetic disfigurement, an increased risk of malignant transformation and neurocutaneous melanosis. The development of melanoma within a giant or large CMN often happens before the age of 5.³ The lifetime risk of melanoma is estimated between 4.5% and 10%. Risk factors include multiple lesions (three or more), large size (> 20cm) and young age (50% arise before the age of three).



Figure 1. Giant congenital nevus



Dr Lynn Chiam is a Consultant Dermatologist at the Children and Adult Skin Hair Laser Clinic at Mount Elizabeth Novena Specialist Medical Centre. She completed her specialist training in dermatology at the National Skin Centre, Singapore. She is an accredited dermatologist and is a Fellow of the Academy of Medicine (Dermatology), Singapore. Dr Lynn Chiam received the Health Manpower Development Programme Award in 2009 for her fellowship in paediatric dermatology at the Department of Dermatology, Radboud University,

Utrecht Medical Centre, the Netherlands.

Infants with neurocutaneous melanosis are rare. These infants are at risk for malignant melanoma of the central nervous system, seizures, and other neurological symptoms due to increased intracranial pressure. Other signs include hydrocephalus, developmental delay and cranial nerve palsies. The risk factors for neurocutaneous melanosis are multiple satellite nevi (> 20) and giant congenital nevus in midline location.

The management of CMN must be individualised. Small and medium-sized CMN can be excised for cosmetic reasons. Large and giant CMN can be removed in part by serial excisions. This surgical debulking theoretically lowers the risk of malignant transformation within the nevus itself but does not eliminate the risk of melanoma completely. The development of rapidly growing ulcerating or atypical-appearing macules, papules, or deep nodules are concerning features and should be biopsied. Close clinical surveillance is paramount for all patients with large/giant CMN.

Café Au Lait Macules

Café au lait macules (CALMs) are hyperpigmented lesions that may vary in colour from light brown to dark brown. This is reflected by the name of the condition, which means "coffee with milk." The borders may be smooth or irregular.

It is a common birthmark and is present at birth in 10%-20% of infants. More than six CALMs over 1.5cm in diameter is a marker for classic Neurofibromatosis type 1 (NF1). In NF1, CALMs tend to increase in size and number throughout childhood. Other signs like neurofibromas, Lisch nodules and axillary freckling do not appear till later childhood/adolescence.

CALMs are also found in the following conditions:

- Neurofibromatosis type 2
- Tuberous sclerosis
- McCune Albright syndrome
- Bloom syndrome
- Ataxia telangiectasia

Nevus Sebaceous

Nevus sebaceous is a common developmental abnormality, classically appearing as a solitary, well-circumscribed, round, oval or linear pink-yellow to orange waxy smooth hairless plaque on the scalp, face or neck. It is a hamartoma comprising sebaceous and apocrine glands. This birthmark is seen in 0.3% of newborns.⁴ Nevus sebaceous tends to enlarge proportionally with the child. During puberty, the lesions can become thicker and more verrucous. In some cases, it can be mistaken for a verruca vulgaris or an epidermal nevus.

Epidermal nevus syndrome is the association of a nevus sebaceous with mental retardation, central nervous system abnormalities, ocular and cardiac defects, and/or skeletal abnormalities.

Prophylactic surgical excision was historically recommended for nevus sebaceous as early studies suggested a relatively high risk (10%) for the development of basal cell carcinoma and other malignancies.⁵ However, recent evidence suggests that trichoblastoma is the most common benign neoplasm to arise within a nevus sebaceous. Recent studies report an incidence of BCC at 0.85%.



Figure 2. Café au lait macules



Figure 3. Nevus sebaceous

Prophylactic surgical excision still remains the treatment of choice for cases requiring aesthetic improvement.

Pigmentary Mosaicism

Pigmentary mosaicism is a general term used to describe any aberrant skin markings that follow a linear, whorled or segmental pattern. This patterned streaking on the skin can be either darker or lighter than the normal background skin. These changes can be seen at birth or within the first few months of life, becoming more pronounced with time before ultimately stabilising. Pigmentary mosaicism is not hereditary.

60% of children affected with extensive pigmentary mosaicism have gross chromosomal abnormalities.²

Infantile Haemangiomas (IH)

Vascular anomalies most frequently present at birth or in early childhood. Of these, infantile haemangioma is most common, affecting up to 10% of infants by one year of age. It is less common in Asians and African Americans. Risk factors include:

- Low birth weight (< 1200g)
- Prematurity
- Female sex (3:1)
- Caucasian ethnicity
- Multiple gestation pregnancy