Congenital melanocytic naevi

What are congenital melanocytic naevi?

Congenital melanocytic naevi (CMN) are skin lesions consisting of nests of melanocytes (cells that produce pigment). They are present at birth or shortly after birth. They occur in approximately 1 in 100 live births.

CMN are classified according to their predicted adult size:

- Small – reach less than 1.5 cm
- Medium – reach between 1.5 cm and 19.9 cm
- Large (giant) – reach at least 20 cm (40 cm)

What causes congenital melanocytic naevi?

CMN usually occur sporadically. The condition is generally not inherited but arises from a mutation in the body's cells that occurs after conception.

NRAS gene mutations cause most cases of giant congenital melanocytic nevus. Rarely, mutations in the BRAF gene are responsible for this condition. The proteins produced from these genes instruct the cell to grow and divide (proliferate) or to mature and take on specialised functions (differentiate). The NRAS or BRAF gene mutations responsible for giant congenital melanocytic nevus are somatic, meaning that they are acquired after conception.

A somatic mutation in one copy of the NRAS or BRAF gene is sufficient to cause this disorder.

These mutations occur early in embryonic development during the growth and division (proliferation) of cells that develop into melanocytes. The overactive protein may contribute to the development of giant congenital melanocytic naevus by allowing cells that develop into melanocytes to grow and divide uncontrollably, starting before birth.

What do congenital melanocytic naevi look like?

Compared to common moles, CMN are often larger and more raised above the skin. Many are different shades of brown and black, and some are hairy.

What other problems can occur with congenital melanocytic naevi?

Due to their appearance, CMN can cause significant psychosocial consequences if they occur on prominent sites.

Giant CMN are associated with a risk of melanoma in the early years and over a lifetime. The risk increases with the size of the lesion and number of lesions. Large lesions (larger than 20 cm and especially larger than 40 cm predicted adult size) are associated with a 2-5% risk of melanoma over a lifetime. The risk of melanoma with small and medium CMN is not significant.

Large or giant lesions have been associated with other cancers.

Another complication of large or multiple lesions is neurocutaneous melanocytosis (melanocytes in the central nervous system). The risk is between 2.5% and 45% depending on factors such as size of the CMN, trunk location and number of satellite lesions. There may be no symptoms. However, a small percentage of children with large CMN and many satellites may experience neurological symptoms such as headaches and seizures.

How are congenital melanocytic naevi diagnosed?

CMN is diagnosed from a clinical examination of the skin. In some cases, a biopsy may be needed.

How are congenital melanocytic naevi treated?

Treatment requires an individualised approach, based on the potential risk factors for complications, psychosocial and cosmetic considerations and the expectations of those affected with this condition and their families.

Infants with large lesions are usually managed by a multidisciplinary team and have regular follow-up by a dermatologist because of the increased risk of complications.

It is important that those with symptoms suggestive, or at high risk, of neurocutaneous melanocytosis have magnetic resonance imaging (MRI) to detect the disease. Those at high risk should have an MRI in the first 6 months of life.

Complete surgical excisions may reduce the risk of melanomas. However, this is associated with complications, and total removal may be impractical for very large lesions.

Non-surgical treatments such as dermabrasion may produce some cosmetic improvement. However, these treatments do not reduce the risk of melanoma as melanomas can occur deep in the skin. Options are best discussed with the dermatologist and plastic surgeon involved.

What is the likely outcome of congenital melanocytic naevi?

CMN usually grow proportionally as the child grows. Some may become lighter with time. However, they generally persist for life.