Epidermal naevus

What is an epidermal naevus (EN)?

It is a term for a group of birthmarks made from cells from the outer portion of the skin (the epidermis), which appear in one or many lines or in a swirled pattern.

What causes it?

Epidermal naevi occur when early in the life of the embryo a genetic mutation develops in one cell, which divides and populates the cells along a line of development (Blaschko's lines). This mutation is not found in other cells of the body.

To date mutations have been found in HRAS, KRAS, PIKC3A, FGFR (fibroblast growth factor 2 and 3), KRT1 and KRT10 (keratin) when the skin is sampled. However, skin biopsy is not the usual way for making the diagnosis.

What does it look like?

Epidermal naevi can have a dark, rough or warty texture (verrucous EN) or a yellow bumpy appearance (naevus sebaceous) or may look like a swirl or line of blackheads (naevus comidonicus) or even can be red and scaly (inflammatory linear verrucous epidermal naevus ILVEN). Becker's naevus usually appears on the trunk of an older child with a raised, brown and hairy patch.

How is it diagnosed?

The diagnosis is usually made by a dermatologist or a paediatrician who has seen the child and recognises the appearance of the lesion(s).

What other problems can occur with EN?

There is an increased risk of hair follicle tumours and rarely skin cancers arising in naevus sebaceous in older children.

Large epidermal naevi may sometimes be associated with malformations in the brain, eyes or bones. The name epidermal naevus syndrome is then used. Rarely bone involvement may result in hypophosphatemic rickets.

EN can be part of other syndromes such as Proteus syndrome.

As the mutation is in the fetal tissue not the parents' sperm or egg, usually there is no risk of having another child with this birthmark. However, there are some situations where the child with the birthmark may be able to pass on the mutation to their offspring. This to date has been reported in some subtypes of EN including epidermolytic EN (keratin 1, 10), linear Darier's (ATP2A2), porokeratotic eccrine naevus (GJB2). This is best discussed with a geneticist. A skin biopsy may help classify some of these types.

How is EN treated?

Small lesions can be surgically excised. Larger lesions should be discussed with a dermatologist and/or plastic surgeon.

What sort of follow-up is needed?

If lumps appear in the lesion, this needs to be reviewed.

If there are associated problems these need to be followed closely.

A skin biopsy in young adult life may be suggested by your dermatologist if there is concern for some of the rare types which can be passed on.