Cutaneous Mosaic Disorders Nevi & Nevoid Skin Disorders and Complex Vascular Malformations and Vascular Tumors

Introduction to DNA and genes

DNA is a code in all living things, including people (Fig. 1). The code is a set of instructions for how to build the living thing, or how to build a person. The instructions are divided up into GENES, which are like words in the code (Fig. 2). We inherit our genes from our parents, and the mixture of those genes makes us who we are (Fig.3).

Fig. 1: DNA helix

Fig. 2: genes are like words in the code

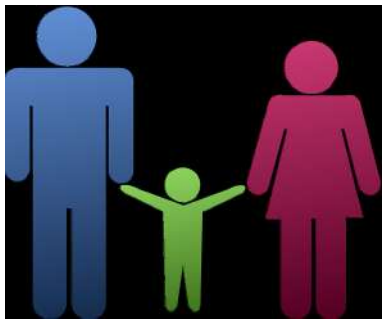

Fig. 3: we inherit our genes from our parents

Introduction to genes and mutations

Mistakes can happen in a gene, like a spelling mistake in any instruction (Fig. 4). A mistake in a gene is called a mutation, that can lead to a change in how the new person is made. In many genetic diseases, a mistake in a gene is inherited from the parents, but in mosaicism the genes from the parents do not carry mistakes (Fig. 5).

Fig. 4: mutations are like spelling mistakes in an instruction

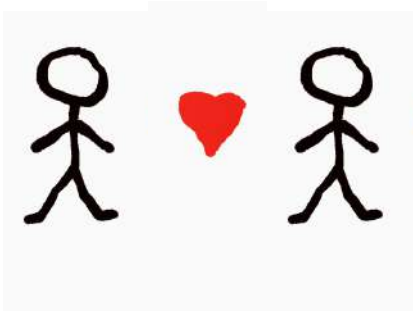

Fig. 5:In mosaicism we do not inherit mistakes our parents

What is mosaicism?

Mosaicism is having a mistake in a gene, which has happened to the baby when it is developing in the womb. The mutation therefore does not affect the whole baby, but just a part of it – like a "mosaic" of two different colours (Fig. 6). When the mutation affects the skin, we see it as a birthmark (Fig. 7).

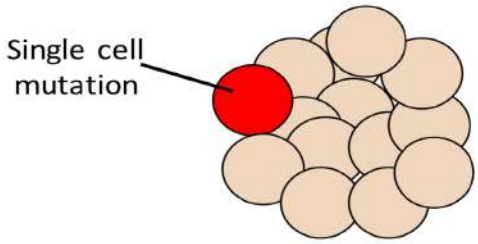

Fig. 6: mutation in a cell of an embryo

Fig. 7: birthmark in embryo due to a mutation

What is the consequence for the patient?

Mosaicism may led to several diseases. They ranges from mild to severe conditions that may involve internal organs (Fig. 8). The principal category of mosaicism are:

a) Vascular anomalies
b) Congenital melanocytic naevi
c) Epidermal nevi
d) Complex syndromes with multiple component

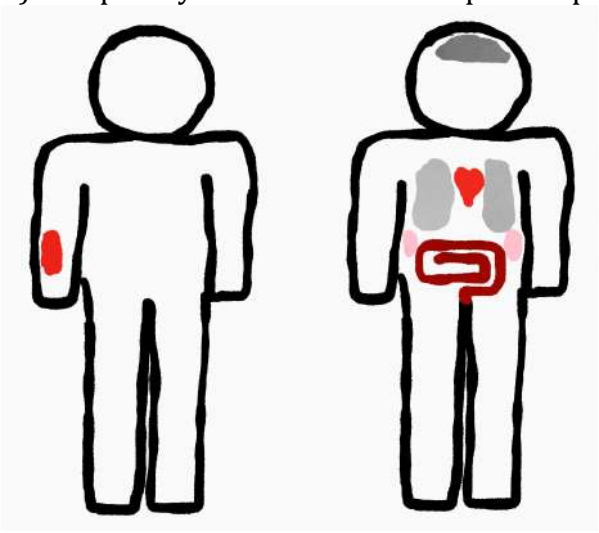

Fig. 8: Consequences may range from a patch on the skin to involvement o f several internal organs.

Is there a risk of passing from the patient to siblings?
In general the risk of passing to siblings is very low and mosaicisms are sporadic (Fig. 9). Ask to your doctor for further information (Fig. 10).

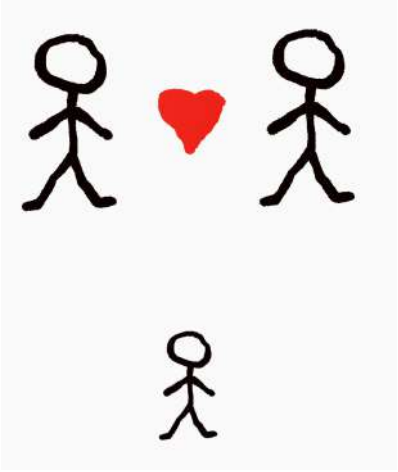

Fig. 9: the risk of passig mutations from a person affected with a mosaic to siblings is negligible

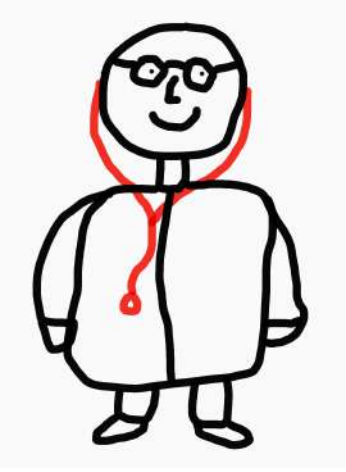

Fig. 10: the Doctor will give you further information on the risk of passing the mutation

The European Reference Networks (ERNs)
The European Reference Networks (ERNs) gather doctors and researchers with high expertise in the fields of rare or low-prevalence and complex diseases. They are "virtual networks" which discuss the diagnosis and the best possible treatment for patients from all over Europe. 24 ERNs were launched in 2017, involving more than 900 highly specialised healthcare teams, located in more than 300 hospitals in 26 European countries.
Among the ERNs, ERN-Skin specifically has the aim to improve the quality, safety and access to highly specialized healthcare providers for rare, low prevalence and undiagnosed skin disorder.

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Cutaneous Mosaic Disorders : Nevi & Nevoid Skin Disorders Complex Vascular Malformations & Vascular Tumors

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European Reference Network
for rare or low prevalence complex diseases
Network Skin Disorders (ERN-Skin)

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