

Additional Genetic Findings in Minors

Information Brochure for Parents\*

\*Updated in April 2024

# What are genetic variants?

Ein Bild, das Frühling, Schraubenfeder, Natur, Typografie enthält.

Automatisch generierte BeschreibungAll externally visible (e.g., hair color) but also invisible characteristics that are passed on via genes are referred to as genetic traits. They are passed on over generations. In the process, changes, new combinations, or deviations in the genes (so-called genetic variants) continuously arise naturally, making each person unique. As we know today, a very small fraction of these variants plays a role in the development of certain (genetically determined) diseases. This small fraction of variants is of particular importance for the person being examined if courses of medical action (e.g., for improved treatment of diseases) can be derived from them. In scientific studies, these variants are also important for researchers because they can contribute to understanding the diagnosis and treatment of diseases. Knowledge about the health significance of different variants is constantly growing. This is leading to improvements in diagnostics and therapies, even though the health significance of many of these variants is currently still unclear due to the large number of possible genetic variants. With medical progress, these will increasingly be uncovered in the future.

# Where do genetic variants come from?

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Automatisch generierte BeschreibungGenetic variants are either **inherited** or **emerge**.

**Ein Bild, das Kunst, Farbigkeit, Screenshot, Design enthält.

Automatisch generierte BeschreibungInherited** genetic variants are passed on from parents to their children. They are already present at the time of birth and continue to exist in a stable manner. If the genetic variants are those for which an association with a disease has already been proven, they can cause symptoms either from birth or later in life. Since inherited genetic variants are passed on from generation to generation, they often allow conclusions to be drawn that blood relatives may also be affected by the variant.

Genetic variants were not inherited in all cases. They may also **emerge** anew from one generation to the next, which is referred to as *“de novo mutation”.*

# Why are genetic variants searched for in the course of diagnostics?

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Automatisch generierte BeschreibungIn **genetic diagnostics**, genetic variants that could be the cause of an already existing disease are searched for. Depending on the findings, **therapy recommendations or preventive options** can be derived from this. Sometimes genetic variants are also searched for in healthy individuals (predictive diagnostics), for example if a family history provides evidence of a genetic disease. Knowledge of such variants is important, for example, in the case of a hereditary predisposition to tumors, so that tumors can be **detected and treated at an early stage**.

# What are additional findings?

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Automatisch generierte BeschreibungIn genetic tests aimed at identifying the genetic cause of a disease and, if necessary, deriving recommendations for its treatment, variants can be discovered that are not related to the original question. These so-called additional findings in genetic diagnostics are thus findings that are discovered in the course of diagnostics, for which no active search was made but which are nevertheless associated with other, possibly inherited and heritable characteristics and diseases.

# How sure can I be that a discovered predisposition will lead to disease?

An additional finding indicates that the **probability of developing a certain disease** is **increased to a greater or lesser extent**. How strong this increase is, depends on the type of additional finding. We will only inform you of additional findings that have a high probability of actually leading to a disease.

# Ein Bild, das Symbol, Kreis, Grafiken, Schrift enthält. Automatisch generierte BeschreibungEin Bild, das Symbol, Pixel enthält. Automatisch generierte BeschreibungEin Bild, das Logo enthält. Automatisch generierte BeschreibungWhat are the types of additional findings?

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Automatisch generierte BeschreibungEin Bild, das Kreis, Kerze enthält.

Automatisch generierte BeschreibungAdditional findings may indicate **predispositions to diseases** for which **preventive programs and/or treatment options** exist, as well as diseases for which (according to current medical knowledge) **no preventive programs and/or treatment options** exist at this time.

Both types of predispositions can lead to disease either in **childhood** or in **adulthood**.

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Automatisch generierte Beschreibung**There are also additional findings that are not medically significant for the person examined because they do not lead to a disease. However, the knowledge of this so-called **disease carrier status** can be significant for their **offspring** and, under certain circumstances, for **parents** and **siblings**.

# What does it mean if no preventive programs and/or treatment options exist at this time?

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Automatisch generierte BeschreibungNew findings are constantly being made** in medical research and the spectrum of diagnosis and treatment is being expanded. Thus, there is a possibility that a currently untreatable disease will be treatable in a few years. However, it is often not possible at the present time to estimate how likely this is to be the case.

# What does it mean that a disease is “treatable”?

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Automatisch generierte Beschreibung*Ein Bild, das Logo enthält.

Automatisch generierte Beschreibung* A disease is considered treatable if **medical measures** areknown that allow this disease to be **prevented** or **therapeutically counteracted**.

# What is a disease carrier status?

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Automatisch generierte Beschreibung**A so-called disease carrier status describes a genetic variant that usually does **not cause any disease in the affected person**. However, the variant can be inherited, which may have consequences for the **offspring**. The probability of disease in the offspring depends on the **mode of inheritance**. For example, in many inheritance modes, the offspring will only become ill (with a probability of 25%) if the partner also happens to carry a disease-causing variant in the same gene (i.e., is also a disease carrier). In other modes of inheritance, a disease carrier status in a female (independent of the partner) is sufficient for the male offspring in particular to develop the disease with a relatively high probability.

# Is there an active search for additional findings?

**No**, additional findings are not actively sought. There is also no obligation to collect them.

# What additional findings will I be notified of?

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Automatisch generierte BeschreibungIn principle, only additional findings are reported that are **highly likely** to lead to a **disease**. The following cases must be distinguished:

Additional findings for diseases for which **preventive programs and/or treatment options** exist that can be carried out in **childhood are always returned** to you.

Ein Bild, das Kreis, Grafiken, Design enthält.

Automatisch generierte BeschreibungIn the case of additional findings for diseases for which **preventive pro- grams and/or treatment options do not exist until** **adulthood**, you can decide whether to

1. receive the additional findings and appropriate genetic counseling.
2. receive information that there is an additional finding that will become medically relevant in your child’s adulthood, without further counseling.
3. receive no return at all.Ein Bild, das Kreis, Grafiken, Symbol, Logo enthält.

   Automatisch generierte Beschreibung

Additional findings for diseases for which there are **no preventive programs and/or treatment options** at this time, and which may occur in **childhood**, will be shared with you **upon request** if they are conditions that may be relevant to your or your child’s **life plans**.

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Automatisch generierte Beschreibung**Ein Bild, das Kreis, Symbol, Grafiken, Logo enthält.

Automatisch generierte BeschreibungAdditional findings for diseases for which there are **no preventive programs and/or treatment options** at this time, and which may occur in **adulthood, will not** be returned to you.

Findings on the **disease carrier status** will be returned to you **upon request** if there is a high probability that you or any siblings can benefit from the return.

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# Can I refuse the information about additional findings?

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Automatisch generierte BeschreibungYou can refuse information on additional findings related to diseases for which there are **preventive programs and/or treatment options in adulthood**, as well as on additional findings for diseases for which there are **no preventive programs and/or treatment options.** You can also refuseto receive information on the **disease carrier status**.

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Automatisch generierte BeschreibungYou **cannot** **refuse** additional findings on diseases that are highly likely to occur in your child and for which **preventive programs and/or treatment options already exist in childhood.** You also cannot refuse knowledge of findings that are **relevant to your child’s current treatment**.

# Ein Bild, das Dunkelheit enthält. Automatisch generierte BeschreibungHow common are genetic variants that are associated with a disease according to current knowledge?

# Studies have shown that if you actively search, you will find genetic variants that are associated with a disease in around 3 out of 100 people who undergo a comprehensive genetic analysis. However, as these studies actively search for these genetic variants, these are not additional findings (but simply findings).

# What are the benefits and potential burdens of receiving additional findings?

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Automatisch generierte Beschreibung**Ein Bild, das Kreis, Symbol enthält.

Automatisch generierte BeschreibungEin Bild, das Schwarz, Reihe, Schwarzweiß, Design enthält.

Automatisch generierte Beschreibung The return of additional findings is only given on the assumption that it is **medically useful** for your child. Nevertheless, the return may also result in **burdens** or **risks** for your child (and possibly also you), such as worry and concern; the need for additional examinations for clarification; insurance aspects; reconsideration of family planning. In addition, a situation may arise in which you have to decide whether to **inform relatives about an identified hereditary predisposition** (which could also affect these relatives themselves) without knowing whether they want to be informed at all. Your doctor can advise you on how to communicate with your relatives.

# What significance can additional findings have for the family?

Genetic predispositions can **emerge spontaneously** (de novo mutation) or be **inherited**. Thus, it is possible that additional genetic findings in your child may also have a significance for **you as parents** or for **siblings**. Consequently, either you yourself may be suffering from **the same genetic predisposition** and be at risk of developing the corresponding disease. Or both parents are **diseases carriers**, i.e., you yourself do not have an increased risk of the disease but have passed the disease on to your child. In both cases, siblingscan potentially also be affected by the same pre- disposition. It is not possible to say with certainty whether you as parents, and therefore possibly also your child’s siblings, are affected by the same predisposition as your child on the basis of the genetic analyses of your child alone. This would require an analysis of your own genetic material (and possibly the genetic material of the child’s siblings).