FOUNDED IN 1988

SWISS PKU

SWISS ASSOCIATION FOR PHENYLKETONURIA AND OTHER INHERITED AMINO ACID METABOLISM DISORDERS

SWISSPKU

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WHAT ARE INHERITED AMINO ACID METABOLISM DISORDERS?

Amino acids are the building blocks of proteins and are an integral part of our food. During digestion, the body breaks proteins down into amino acids, which are then absorbed by the body or further processed by enzymes.

In rare cases, inherited genetic differences can result in certain enzymes being deficient or lacking. This means that the enzyme cannot break down its corresponding amino acid, or can only break it down incompletely. If this situation is left untreated, the particular amino acid builds up in the blood to a level that is multiple times the usual concentration. This can lead to serious physical and mental health problems. To prevent disability, it is important that any metabolic disorders are identified as soon as possible after birth. This is why, since 1965, all newborns in Switzerland have been screened in the first few days of life to test for the main metabolic disorders.

There are a number of different amino acid metabolism disorders affecting different enzymes and amino acids. They are treated by giving the affected children a low-protein diet combined with a special protein powder to ensure their diet does not lead to protein deficiency. Specific medications are also used to treat some metabolic disorders. With early detection and a strict diet, children affected can live healthy lives despite having an amino acid metabolism disorder.

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THE MAIN AMINO ACID METABOLISM DISORDERS

Phenylketonuria (PKU)
People with the inherited amino acid metabolism disorder PKU cannot break down the amino acid phenylalanine or can break it down only partially. This leads to an accumulation of phenylalanine in the body. If left untreated, this interferes with brain maturation and causes severe, irreversible physical and mental health problems.

This can be prevented with a low-protein diet to control the intake of phenylalanine. The diet is supplemented with a phenylalanine-free protein powder to ensure daily protein intake is sufficient. The diet is followed under medical supervision and the person affected is supported by a dietician. Progress is monitored with regular blood tests. If they adhere strictly to the special PKU diet, children with this disorder can live healthy lives with no physical or mental problems.

Treatment with the medication sapropterin may allow the strict diet to be relaxed. Around 30% of people with PKU respond to this treatment.

Maple syrup urine disease (MSUD)
In MSUD the amino acids leucine, isoleucine, and valine cannot be broken down or are broken down only partially. If the person's diet includes normal amounts of protein, these amino acids will accumulate in the body to a dangerous level, potentially leading to brain damage and death.

Treatment consists of a low-protein diet supplemented with a special amino-acid mixture to ensure daily protein intake is sufficient.

Tyrosinaemia

Tyrosinaemia describes a group of inherited amino acid metabolism disorders that affect the way the amino acid tyrosine is broken down in the body. The process of breaking down this amino acid is blocked, generating toxic by-products. Left untreated, these disorders lead to damage to the brain and other organs and sometimes even death. Treatment can vary depending on the type of tyrosinaemia, but usually consists of a low-protein diet supplemented with a specific amino-acid mixture to ensure daily protein intake is sufficient. Tyrosinaemia type 1 is also treated with medication (NTBC).

Homocystinuria

In the inherited amino acid metabolism disorder homocystinuria, the amino acid methionine is not converted into cysteine, but instead is converted into the harmful amino acid homocysteine. High levels of homocysteine in the blood can cause a number of serious symptoms relating to various organs (e.g., cardiovascular disease, developmental delays). These complications can be prevented with a low-methionine or low-protein diet. Protein requirements are met by supplementing the diet with a methionine-free amino-acid mixture. Treatment may also include betaine and specific vitamins.

Glutaric aciduria type 1

Glutaric aciduria type 1 is caused by a genetic defect that results in an inability to properly break down the amino acids lysine and tryptophan, which then accumulate in the body. Levels of harmful by-products build up and can cause irreversible brain damage. Disability can be avoided through adherence to a special low-protein diet and specific amino-acid supplementation, coupled with prompt treatment with intravenous fluids if the person develops an infection. The medication carnitine is used to treat this disorder.

Isovaleric acidaemia

Isovaleric acidaemia is an inherited amino acid metabolism disorder in which the amino acid leucine is not broken down properly. Instead, isovaleric acid builds up in the body and causes damage to the brain and nervous system. A low-protein diet is followed to restrict leucine intake. The diet may need to be supplemented with a leucine-free amino-acid mixture. The medications carnitine and glycine are used to treat this disorder.

Methylmalonic and propionic acidaemia

These diseases are inherited disorders of propionate and methylmalonate metabolism, which in healthy humans serves to generate energy from food proteins. In people affected by these disorders, the amino acids methionine, threonine, valine and isoleucine are not properly utilised. Instead, toxic metabolic products accumulate in the blood and can cause serious damage to the brain and other organs. Damage can be prevented by following a strict low-protein diet. To meet protein requirements, the diet is supplemented with an amino-acid mixture that contains all necessary protein components except methionine, threonine, valine and isoleucine. Some patients can be treated with the medications carnitine and vitamin B12.

Urea cycle disorders

The urea cycle occurs in the cells of the liver and converts nitrogen, which is obtained from proteins in food, into urea. In some inherited genetic conditions, this conversion may be disturbed, causing toxic ammonia to build up in the blood. If left untreated, this build-up, known as hyperammonaemia, can lead to brain damage and even death. However, if it is detected early, the ammonia concentration in the blood can be lowered with medication and a low-protein diet. If the person follows this diet strictly, permanent damage can be avoided.

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WHAT DOES SWISS PKU DO?

Swiss PKU (formerly CHIP) is a Swiss association for people with phenylketonuria (PKU) and other amino acid metabolism disorders. It was founded in September 1988 by a group of dedicated parents and now has more than 130 members.

Our main aims are to support the quality of life of those affected and create the best possible conditions so that people can live an independent life despite having an amino acid metabolism disorder.

To achieve these goals, we:

- organise a variety of events for people affected (e.g. a children's camp and cooking classes),
- encourage and facilitate the exchange of information and experience between members, professionals and the general public,
- hold an annual general meeting with relevant specialist presentations, and
- support the importation of special low-protein foods to Switzerland.

Swiss PKU is a member of the European Society for Phenylketonuria E.S.PKU (www.espku.org).

More information about inherited amino acid metabolism disorders, Swiss PKU and current events can be found on our website: www.swisspku.ch

PKU and special diets

'Beratungsstelle Oase' is an advisory centre affiliated to Swiss PKU. The centre dietician, Cäcilia Smith, helps families who have questions about low-protein diets and offers valuable support and lots of practical tips.

Phone +41(0)79 646 66 10
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WHO CAN I CONTACT FOR MORE INFORMATION?

More about Swiss PKU, current events and useful information can be found on our website: www.swisspku.ch

To join the association or find out more:

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