

# Living with Phenylketonuria

**Scene no.1 – Don Juan (Karel) goes back to the restaurant. The waiter is bringing penne with chicken meat, cream and broccoli. Just behind him is another waiter with an apple and carrot salad and chips with ketchup.**

While Juan (Karel) is enjoying his pasta and his girlfriend Adélka is slowly chewing away one chip after another, they are making wedding arrangements. This is how looks the luckier variant of the life with phenylketonuria, a disease that does not allow you to eat anything (or just the bare minimum) of what contains proteins.

Today twelve-year-old Adélka from Česká Třebová does not know yet what direction her life will take. But she is sure that her life will be far from easy. A few days after her birth, just as all babies do by the time they are one week old, she had her phenylketonuria tests done and this was how her mother learnt that Adélka's life will be complicated by this disease which entail a very poor diet.



The first five years, the strict diet was not a problem. Adélka's mum cooked her food, sometimes her grandmothers helped as well. They all made sure that the child did not exceed the daily protein allowance and that, at the same time, she had all the important nutrients of the phenylketonuria diet: a special drink rich in amino-acids, microelements and vitamins – which the body lacks when it has to follow such a strict diet.

Even when Adélka started school, it did not seem at first, that she would be too limited by her „eating handicap“. The classes in the first and second year finished at lunchtime and just as all her classmates, Adélka brought her snack from home. And she simply did not take lunch at school. She did not have to. She could go straight home.

## School, the basis of life

However, as she was getting older, afternoon classes started as well. Adélka's journey to school takes forty minutes by public transport, so she did not have time to go home for lunch. And this is when the problems started.

Her parents asked the school, if Adélka could get phenylketonuria lunch at least twice a week, but the school refused. Therefore they announced the direction that Adélka would bring her own food into the canteen. However, the reply they received said, that on sanitary grounds, it is not possible in the canteen to heat up food brought in from the outside. What is more, it was insinuated to us that the best would be if Adélka did not bring any food at all in the canteen, her mother recalls the difficulties they had three years ago.

In the end, the school offered a seemingly illogical solution. Adélka can eat her food in the corridor. According to some regulations, a teacher is responsible for pupils only in the classroom, not in the corridor. This way, the school could easily get away with any complications that might come up in case something happened to Adélka while she eats her lunch; Adélka's mum describes the situation. However, she did not like the idea at all, that Adélka should have her lunch in the corridor. Therefore she signed a declaration that she assumes all the responsibility for her daughter during and outside classes and the problem was sorted out.

## Where is the humanity in all that?

Leaving aside the fact that Adélka's mother has to cook a special meal every day from ingredients that are often hard to find, one question arises: Adélka now brings her own snack and lunch to school. She can calmly and with some dignity have her food at her desk. But is not there something else missing?

Try to remember, how many great friendships started around a spaghetti battle in the school canteen. And the number of unforgettable loves, which, but for the cheap black tea with lemon, would be impossible? We know that Adélka will always stand aside with her different eating needs. But to isolate a child from a natural collective of children is, to say the least, beyond all

understanding. We know that there are regulations that the school does not want to and cannot break. Yet we do not understand why such regulations are made, Adélka's mum shrugs her shoulders.

## Detection and threat

### Diagnostics

Phenylketonuria is detected via screening of new born babies between the third and eighth day after the baby is born. A drop of blood is taken from the baby's heel and placed onto a test card, which is sent to a special laboratory. In the Czech Republic all new born babies are tested this way. If the tests are positive or borderline, parents are immediately informed and an appointment is arranged with the advisory centre for metabolic disorders where the doctor together with the parents determines further procedure.

Phenylketonuria is a genetic disease, this is why, in order to diagnose it, and it is possible to make use of a so called pregnancy screening. This is used in case one of the parents or somebody in the family has or had the disease. The probability of phenylketonuria is said to be 1:10 000 according to some sources, and 1:8000 according to others. Every year, there are approximately ten cases of phenylketonuria diagnosed in our country.

## Phenylketonurian and Mental Damage

Phenylalanin is an amino-acid found in all proteins. While a healthy organism processes this substance – it splits it and copes with any amount of it, a sick body cannot make use of it nor can it excrete it properly. As a result, phenylalanine accumulates in the body until its high concentration starts to damage cells.

Unless the disease is detected and treated in time, the development of the nervous system quickly becomes damaged, i.e. it leads to mental retardation (microcephalia) and slows down psychomotor development. People suffering from phenylketonuria often have blond hair and blue eyes. Their urine has a specific odour (so called mice odour). It is very important that a child follows a diet from the moment of detection of phenylketonuria until the completion of the development of brain. Even when adults, the diet does not end, mainly for women who wish to have babies. They have to be very careful about what they eat, so that the development of the baby is not endangered. If adults do not keep the diet, there is a risk of a direct link with Alzheimer disease.