

What do we know about how high phe levels affect adults?

Are you wondering what we know about PKU in adults? Here are the results of a recent study that was published in the Journal of Inherited Metabolic Disease called “Phenylketonuria in adulthood: A collaborative study”. (volume 25 #5 pp 333-346)

This was a follow-up study. The original study was done from 1967-1983, and it followed babies with PKU that were given a low phe diet until age 6 and then were either put in a group to continue diet or to stop diet and formula all together. The original study showed that participants who had continued on the diet developed substantially better than those who discontinued the diet at age 6. These findings were one of the main reasons that medical clinics started recommending ‘diet for life’. Despite recommendations for patients to stay on diet, some of the adults in the original study stopped the diet, so this follow-up study was done to look at these same patients as adults.

In 1998, 70 of the original study participants (who were now adults) were found and enrolled in a follow-up study. Below are the results.

Part One: Medical results

The medical evaluations show that the subjects who stayed on a low phe diet had fewer problems than those who stopped. If you stopped the diet you were more likely to have:

- eczema (28%)
- asthma (12%)
- mental disorders (41%) – these include phobias and depression
- headaches (31%)
- hyperactivity/being busy (14%)
- hypoactivity/tiredness (19%)

Part Two: Genotyping

Genotyping was done in a smaller group of the study participants. PKU is a genetic disorder. This means that a person has the disorder from the time they are conceived. At conception, a baby receives two sets of genetic material, one from the mother and one from the father. This genetic material, called DNA, acts as a recipe for the baby’s development. Each pair of genes gives directions to a certain part of the body. Scientists have found several hundred mutations (mistakes) that can occur in the Phenylalanine Hydroxylase gene. Mutations in this gene can result in PKU. Genotyping is when scientists look at your DNA to find out what kind of mutation you have in your genes – in this case they were looking specifically at the Phenylalanine Hydroxylase gene. In this study 60% of patients had classical PKU by genotype.

Of this group with classical PKU:

-15% had never discontinued diet/formula and their mean adult IQ was significantly higher than the mean adult IQ of the 85% who discontinued diet at an average age of 8 years.

-of the 40% that re-started diet after stopping for awhile, 9 were still taking the medical food and showed an increase in adult IQ compared to childhood IQ

-in contrast, the 18% who did not manage to continue the diet experienced a drop in adult IQ compared to childhood IQ

-Individuals with classical PKU currently taking the medical food had significantly higher adult IQ scores than those on a regular diet

Part Three: Psychological testing

Tests showed that people with higher childhood/adult blood phe levels who had stopped the diet had lower intellectual and achievement test scores.

Part Four: MRI/MRS testing

In a smaller group of cases, Magnetic Resonance Imaging and Spectroscopy (MRI/MRS) were done. This is a way to study brain phe concentrations. 22 people were selected for this substudy and they were split into 2 groups.

Group A: these people were on the phe-restricted diet from birth until at least 10 years of age, and were currently taking the formula as the main source of protein in their diet.

Group B: consisted of 12 people who had discontinued the low phe diet by age 10 years (mean age 6.8 years).

Brain concentrations of phe were measured as well as *blood* phe levels. Average brain phe concentrations were about 40% higher in group B (the off diet group) compared to group A.

Group A had higher IQ's than group B. People in group B (who had higher brain phe levels) had more abnormal MRI results.

The Bottom Line is:

Stay on diet! You will be less likely to have eczema, asthma, mental disorders, headaches, hyperactivity, hypoactivity/tiredness, or abnormal MRI results. People who have never discontinued diet have higher IQ's, intellectual and achievement scores than those people who have stopped the diet. And it is not too late! – if you discontinued your diet and you start back on it you may see an improvement in some of the symptoms above as well as an improvement in IQ level!