

UNDERSTANDING GYRATE ATROPHY

What is gyrate atrophy?

Gyrate atrophy is an inborn error of metabolism affecting the **choroid** and the **retina** of the eye. People with gyrate atrophy gradually lose cells (atrophy) in the retina, the specialized light-sensitive tissue that lines the back of the eye, and in a nearby tissue called the choroid.

How is gyrate atrophy caused?

Gyrate atrophy is genetic. This means that the 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. DNA includes information about the baby's eye and hair colour, sex, and even whether the baby will be right or left-handed. DNA comes in units called **genes**. Each pair of genes gives directions to a certain part of the body.

Genes give instructions to the body on how to break down protein. Everyone needs protein, found in meats, dairy products, fish, eggs, nuts and other foods. The body uses protein to build new tissue and to repair damaged tissue. When someone eats food that contains protein, the body uses what it needs by breaking it down into **amino acids** (the building blocks of protein). Amino acids are used by the body to build skin, muscle, blood and other tissues. Arginine and ornithine are 2 amino acids.

Gyrate atrophy is caused by getting **two copies** of the faulty gene, one from the mother and one from the father. If the child inherits only one copy of the gene, they are a **carrier** for that gene but are not affected by the disease because they make enough enzyme to stay healthy.

What happens in gyrate atrophy?

In gyrate atrophy, an enzyme called **ornithine ketoacid aminotransferase** or **OAT** is missing. OAT's job is to break down the amino acid ornithine. Ornithine therefore builds up in the blood and causes damage to the thin coating of the eyes, the **choroid**, and the **retina**. This results in a gradual

loss of vision, beginning with nearsightedness, loss of night vision and peripheral vision in childhood. As time goes on, people with gyrate atrophy develop tunnel vision and many develop cataracts. These visual changes lead to total blindness between the ages of 40 and 60.

How is gyrate atrophy treated?

The first part of treatment is **reducing protein in the diet**. If less protein is taken in, there is less arginine for the body to change to ornithine:



(Ornithine does not exist in the food we eat, it is made in the body)

Protein in the diet is lowered by avoiding protein-rich foods like meat and milk. Examples of low protein foods are fruits, vegetables and starches. These foods provide calories without loading the body with protein.

Calories are a very important part of the diet. The body can use them for fuel without breaking down its own reserves. If the body does not have adequate fuel in the form of calories, it will actually begin to break down muscle to provide energy. Muscle is protein and will increase ornithine levels in the same way as eating protein will.

The metabolic dietitian works with people with gyrate atrophy to make adjustments to diet. This ensures that adequate calories and nutrients are being provided. It is important for the person to keep a record of what they eat on a daily basis.

The amount of food and drink the person can safely consume is calculated by the dietitian. All foods must be carefully measured to control the amount of protein consumed.

Examples of foods that must be eliminated from a low-protein diet are:

- Milk and dairy products
- Meat, fish, chicken
- Nuts (including peanut butter)
- Beans
- Eggs

It is not unusual for someone on a low-protein diet to have 2 kinds of vegetables and a baked potato for dinner. However, if these foods were all that the person ate, their diet would be lacking in protein, vitamins and minerals. That is where the special medical formula comes in. It provides all the protein and nutrients normally obtained from high-protein food, but is missing the amino acid, arginine. In order to slow down the disease progression.

Some people with gyrate atrophy are helped by **pyridoxine** or **vitamin B6**. These people are said to be "**pyridoxine responsive**" and do not need any other special treatment. Only about 5% of people with gyrate atrophy are pyridoxine responsive. All patients with gyrate atrophy are given a trial of pyridoxine to see if they are responsive.

Useful resources:

Genetics Home Reference

Gyrate Atrophy of the choroid and retina

<http://ghr.nlm.nih.gov/condition/gyrate-atrophy-of-the-choroid-and-retina>