What is GA1?

GA1 stands for Glutaric Aciduria Type 1

It is an inherited metabolic condition
Too much glutaric acid in the blood

Too much glutaric acid in the urine

What is GA1?
GA1 and protein

GA1 affects the way your baby breaks down protein

Many foods contain protein

The body needs protein for growth and repair
What is protein?
Protein and enzymes

Protein is broken down into amino acids (building blocks of protein) by enzymes (which are like chemical scissors).

Enzymes then further break the amino acids into smaller parts.
Protein metabolism

Metabolism refers to the chemical processes that occur inside the cells of the body.
What happens in GA1?

In GA1, the body lacks an enzyme called glutaryl-CoA dehydrogenase.

This means that the body is unable to break down two amino acids called lysine and tryptophan. This leads to a build-up of glutaric acid.
What can go wrong in GA1?

The basal ganglia in the brain controls movement.

The build up of glutaric acid damages the basal ganglia and causes movement problems.
Metabolic crisis

- A **metabolic crisis** can trigger the **movement problems**. This is because there is a build-up of glutaric acid and other toxic chemicals.

- It is usually caused by childhood infections or viruses causing high temperatures, vomiting and diarrhoea.

- Avoidance of a metabolic crisis is essential.
What about other symptoms in GA1?

Babies with GA1 are usually healthy at birth although many are born with a larger than average head size.
How is GA1 diagnosed?

GA1 is diagnosed by newborn screening. High levels of glutaric acid are found in the blood.
How is GA1 managed day to day?

GA1 is managed with the following special diet and medication:

A protein restricted diet

A protein substitute

Carnitine medication
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Protein balance is needed in GA1

In GA1, it is important that enough protein is given for growth...but not too much as toxic chemicals will be made.
Measured protein intake

In babies, a restricted amount of protein is given from breast milk or measured amounts of infant formula. The amount given will be monitored regularly by your specialist metabolic dietitian.
Protein substitute

Protein substitute is essential for metabolic control.

It will help to meet your baby’s protein, energy, vitamin and mineral requirements.

It is available on prescription.
How is GA1 managed during illness?

• During any childhood illness, an emergency regimen is given
• Illnesses can cause catabolism or protein breakdown
• This will lead to a build up of glutaric acid and cause a metabolic crisis
How is GA1 managed during illness?

Stop all protein in food & drink

Start the emergency regimen. This is made up of protein substitute and glucose polymer

Carnitine medication as prescribed

Phone your metabolic team
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Phone your metabolic team
Checklist for illness

Always take full amounts of emergency feeds as prescribed

If symptoms continue and/or you are worried, go immediately to the hospital

Regularly update your metabolic team
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Regularly update your metabolic team
It is imperative that emergency feeds are started **promptly** and there are **no delays** in management.
How is GA1 monitored?

Frequent blood tests to check amino acid and nutrient levels

Height and weight

Developmental checks

Diet and medications are adjusted according to age, weight and blood chemical levels
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Humans have chromosomes composed of DNA.

Genes are pieces of DNA that carry the genetic instruction. Each chromosome may have several thousand genes.

The word mutation means a change or error in the genetic instruction.

We inherit particular chromosomes from the egg of the mother and sperm of the father.

The genes on those chromosomes carry the instruction that determines characteristics, which are a combination of the parents.
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Chromosomes, genes, mutations
Inheritance

GA1 is an inherited condition. There is nothing that could have been done to prevent your baby from having GA1.

Everyone has a pair of genes that make the glutaryl-CoA dehydrogenase enzyme. In children with GA1, neither of these genes works correctly. These children inherit one non-working GA1 gene from each parent.

Parents of children with GA1 are carriers of the condition.

Carriers do not have GA1 because the other gene of this pair is working correctly.
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Inheritance – Autosomal recessive (carriers of GA1)

Mother is a carrier of GA1

Father is a carrier of GA1

Female egg cells

Male sperm cells
Inheritance  – Autosomal recessive – possible combinations

Mother is a carrier of GA1

Father is a carrier of GA1

Child will not be a carrier of GA1

Child will be a carrier of GA1

Child will have GA1

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When both parents are carriers, in each pregnancy the risk to the baby is as follows:

- 25% chance (1 in 4) of GA1
- 50% chance (1 in 2) for the baby to be a carrier of GA1
- 25% chance (1 in 4) for the baby to have two working genes and neither have GA1 or be a carrier
GA1 is a serious inherited metabolic disorder that can lead to severe movement problems

Children are very vulnerable in the first 6 years of life

Damage can be prevented with a protein restricted diet, a protein substitute and carnitine

Remember, during illness, it is imperative that emergency feeds are started promptly, followed strictly and there are no delays in management

With good management, severe movement problems can be prevented in the majority of children
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Take home messages

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- Children are very vulnerable in the first 6 years of life
- Damage can be prevented with a protein restricted diet, a protein substitute and carnitine
- Remember, during illness, it is imperative that emergency feeds are started promptly, followed strictly and there are no delays in management
- With good management, severe movement problems can be prevented in the majority of children

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Always ensure you have a good supply of your dietary products and medicines and that they are in date.

Your dietary products and medications are prescribed by your GP. These are obtained via a pharmacy or home delivery.

Always ensure you have your emergency feed products and a written emergency plan.

Medications to control fever should be given as normally recommended – always keep supplies available.
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Who’s who

- My dietitians
- My nurses
- My doctors
  - Contact details, address, photos
Visit www.lowproteinconnect.com and register to get access to support and practical advice for those living on a low protein diet.

The site also provides information on upcoming events and personal stories from others on a low protein diet.