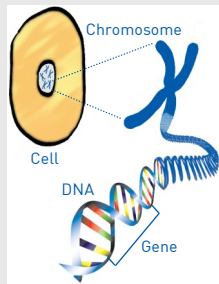


Why did this happen?

Our bodies are made up of millions of cells, which arise from one single cell made at the time of conception. Each cell contains 46 chromosomes, arranged in 23 pairs, on which genes are arranged like beads on a string. Genes act as instructions telling our bodies how to grow, develop and function. Since chromosomes come in pairs, genes also come in pairs. Each parent passes on only one copy of each pair of chromosomes so that the baby has a new set of pairs of chromosomes inherited equally from father and mother.



Occasionally, the genetic code of each parent of the child contains one faulty copy of a gene. In the case of Alazami syndrome, this is the *LARP7* gene. When each parent has one faulty copy and one normal, functioning copy of *LARP7*, they are said to be a “carrier” for the condition and show no features of Alazami syndrome. Alazami syndrome occurs when a child inherits the faulty copy of the *LARP7* gene from each parent meaning they have two faulty copies. Therefore, Alazami syndrome is known as a “recessive” condition.

Can it happen again?

If both parents are carriers for the same recessive condition, there is a 1 in 4 (25%) chance that any child will inherit the faulty gene from each parent and therefore inherit the condition. Conversely, there is a 3 in 4 (75%) chance that any child will be healthy.

A clinical geneticist can give you specific advice for your family and, if applicable, discuss options for testing regarding future pregnancies.

Families say ...

“ You really can't be miserable around her, she's far too happy (99% of the time!). She has enriched our lives as a family and reminds us all that even in dark times and hard times there's always an opportunity to dance and be happy, making us all better people by taking time to recognize her differences, embrace them and value her uniqueness. ”

Websites, Facebook groups and other links

There is a FB group for Alazami Syndrome at:
www.facebook.com/alazamisynndrome/

Inform Network Support



Rare Chromosome Disorder Support Group,

The Stables, Station Road West, Oxted, Surrey RH8 9EE, UK
Tel/Fax: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

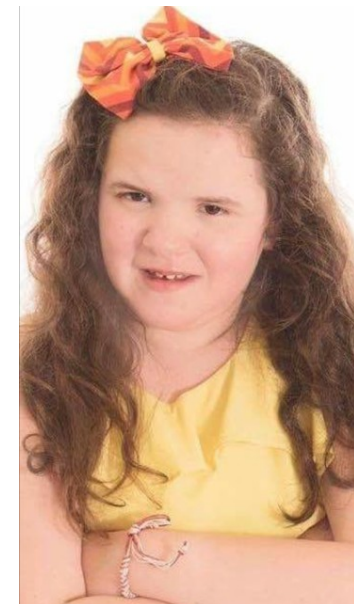
This guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. Unique does its best to keep abreast of changing information and to review its published guides as needed. This guide was written by Dr. Kate Chandler (Consultant Clinical Geneticist) and Dr Zerine Hyder at St Marys Hospital, Manchester 2018 Version 1 (CA)

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Alazami Syndrome
rarechromo.org



What is Alazami Syndrome and what causes it?

Alazami Syndrome, named after the person who first described it, is a very rare genetic condition that arises from changes in both copies of the gene *LARP7*. Only six families with children with Alazami syndrome have been reported to date. The exact function of the *LARP7* gene is not well understood, but it is thought to be involved in maintaining the structures found at the ends of our chromosomes.



“ She was a small, tiny child until age 11 when she suddenly grew and gained weight. At 13 she is average height and slightly overweight! ”

Most Common Features:

- Severe growth delay and short stature
- Autistic behavioural traits
- Global developmental delay and intellectual disability, which is usually severe
- Small head size (microcephaly)
- Triangular-shaped face
- Prominent forehead with deep-set eyes and broad nose
- Wide mouth with widely-spaced teeth

Not all of these features will be present in every affected individual and the way that people are affected can range from a mild to more severe presentation.

Medical concerns

■ Ataxia

Ataxia, which is a lack of voluntary coordination of muscle movements, has been reported in a few children. This can often manifest as a clumsy walking pattern, with frequent trips or falls.

■ Seizures

Seizures have been reported in Alazami syndrome. One person had febrile convulsions as a child, which is when a seizure occurs during episodes of high temperature, but did not go on to develop epilepsy.

■ Thickened skin

Thickened dry, wrinkled skin over the hands and feet is a common feature of Alazami Syndrome.

■ Strabismus

Strabismus is a condition in which the eyes do not properly align with each other when looking at an object and has been reported in a few individuals with Alazami syndrome. Some of the most common ways that strabismus can be treated include special glasses or an eye patch.

■ Heart Problems

Heart defects including atrial septal defects (a hole between the top chambers of the heart) and pulmonary stenosis (narrowing of the heart valve) have been reported in some children with Alazami syndrome. Many heart defects may heal spontaneously, while others may need additional treatment, sometimes including surgery.

Management:

- A paediatric neurologist will be involved in managing children with seizures or ataxia, to tailor medication appropriately.
- A specialist eye team monitors strabismus.
- Children with heart defects should be reviewed by a paediatric cardiologist, and have regular echocardiograms.

Development

■ Growth

Some babies have restricted growth in the womb, and the majority are born with a low birth weight. Babies with Alazami syndrome usually struggle to gain weight, and grow slowly, with ultimate short stature. However, there are a few reports of children with no growth problems at all. A few children have microcephaly, which is a relatively small head size.

■ Physical development

Most children are delayed in reaching developmental milestones such as sitting, crawling and walking.

■ Speech

The majority of children will have a delay in speech and language development, which can be severe. Some children may not develop speech. Communication with other methods including sign language and gestures is often used.

■ Learning

Most children need considerable support with their learning. They may need to attend a special school to receive appropriate academic support, and may require input with their non-academic and daily living skills. Most adults will require long-term supervision.

■ Behaviour

Many children do not have any specific behavioural problems. However, some are reported to display anxious, repetitive or self-stimulating behaviour, including autistic traits.

Management:

- Community paediatricians oversee the care of children with Alazami syndrome, and monitor their development and behaviour. They often coordinate the appropriate specialist teams, including physiotherapy; occupational therapy; and speech and behavioural therapies.
- Children with growth delay should be referred to a paediatric endocrinologist, for consideration for growth hormone treatment.