UNIQUE

UNDERSTANDING GENES & CHROMOSOMES

Perrault syndrome

rarechromo.org

This guide is designed to help people with Perrault syndrome and their families, and the healthcare professionals looking after them. It contains information about the cause, the ways in which it can affect people and suggestions about the help and management that can benefit people with this condition.

What is Perrault syndrome?

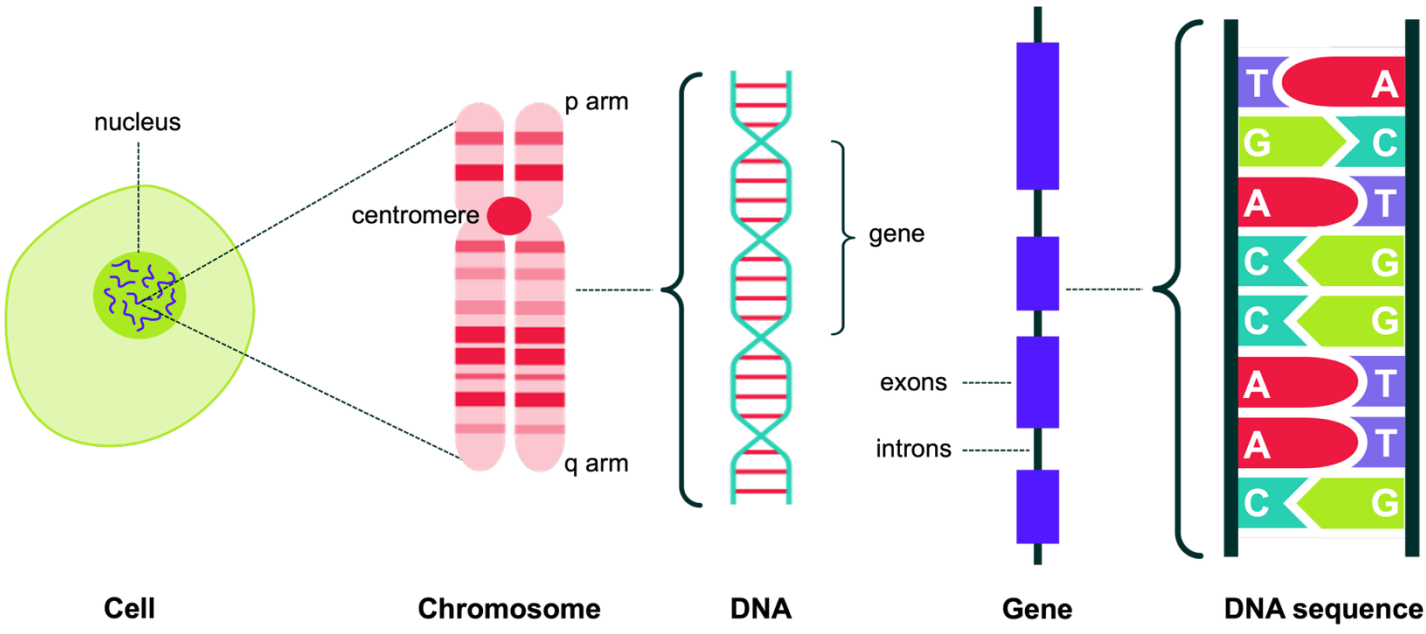
Perrault syndrome is a rare genetic condition. There are two types of Perrault Syndrome. Type 1 Perrault syndrome was first reported in 1951 when Perrault syndrome was thought to only affect females. This milder form of Perrault syndrome is characterised by hearing loss in both ears resulting from damage to the auditory nerve or inner ear (cochlea) (bilateral sensorineural hearing loss (SNHL)), and ovaries that don't function as expected (ovarian dysfunction). Subsequent advances in genetic testing led to the discovery that males could also be affected and also that in many people a wider range of features are observed, including those that affect development, learning and movement. This form of Perrault syndrome, where additional features are also present, is called Type 2 Perrault syndrome.

As is common with genetic conditions, each person is affected differently - even among affected members within the same family.

What causes Perrault syndrome?

Genes are instructions that have important roles in our growth and development. They are made of DNA and are incorporated into organised structures called chromosomes. Chromosomes therefore contain our genetic information. Chromosomes are located inside our cells, the building blocks of our bodies. In people with genetic conditions, one or more of their genes don't instruct the body as we would expect, which can lead to changes in how their body works.

DNA is made up of building blocks called 'bases' or 'nucleotides'. There are four DNA bases which can be abbreviated to the letters A, C, G, and T. These DNA bases are paired up in the DNA structure into 'base-pairs'. The full sequence of our DNA is over three billion base-pairs long. There are changes in the DNA sequence (variants) present in everyone's genes. It is variants in our genes that make each one of us unique individuals.



Most genetic conditions are caused by changes to genes that provide instructions for making proteins; these are called protein-coding genes. Proteins are molecules that are made up of long chains of chemicals called amino acids. Proteins play many critical roles in the body including in processes such as breaking down food, moving our muscles, and growing the body's organs and making sure they function properly.

When a genetic change causes proteins to be built incorrectly, this can lead to a change(s) in the way the body functions, which causes the features and symptoms of that particular rare genetic condition, in this case Perrault syndrome.

To-date (2025), the diagnosis in approximately 80 percent (8 in 10) of individuals with Perrault syndrome has been found to be caused by specific changes (known as pathogenic variants) in the DNA sequence of one of 12 genes: *CLPP*, *DAP3*, *ERAL1*, *GGPS1*, *HARS2*, *HSD17B4*, *LARS2*, *MRPL49*, *PEX6*, *PRORP*, *RMND1* and *TWNK*.

We have two copies of each chromosome, so we also have two copies of each of these genes. Perrault syndrome occurs when both copies of one of these 12 genes are affected. Perrault syndrome is an autosomal recessive condition since all numbered chromosomes are called autosomes and genetic conditions that only occur when both copies of an autosomal gene are affected are known as recessive.

Unique publishes a separate guide to single gene disorders – autosomal recessive

Genetic Test Results

The results of genetic (genomic) testing are likely to be given to you by your geneticist, a genetic counsellor or a specialist/provider who ordered the test e.g. an audiologist.

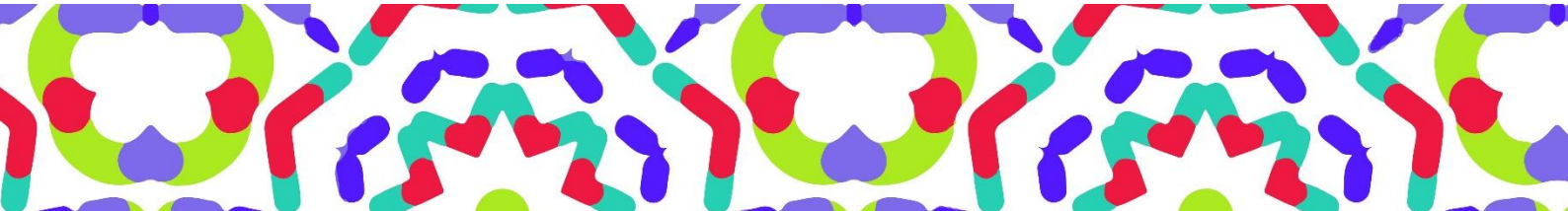
An example result of a DNA sequencing test (e.g. whole exome sequencing (WES) or whole genome sequencing (WGS)), which can identify gene variants, is shown below for the *LARS2* gene.

p.Asn153His c.457A>C in exon 6 of the LARS2 gene (NM_015340.3)

p.Asn153His	Signifies the change to the protein: the amino acid asparagine (Asn also called N) has been replaced/substituted by the amino acid histidine (His also called H) at position 153 in the sequence of amino acids that make up the protein. This is predicted to alter the function of the resulting LARS2 protein.
A>C	signifies the gene sequence change; the A nucleotide has been replaced by a C nucleotide
c.457	signifies the base pair position of the change within the gene sequence (the position where the A nucleotide has been replaced by the C nucleotide)
exon 6	signifies which part of the gene has been altered, in this case exon 6
LARS2 gene	signifies the gene that is affected
NM	denotes the reference sequence used.

NB Another pathogenic variant in this individual's other copy of the *LARS2* gene would also be present.

Unique publishes separate guides to DNA sequencing and Interpreting Genetic Test Results



What features and symptoms do people with Perrault syndrome have?

As is common with many genetic conditions, children and adults with Perrault syndrome can have a range of symptoms and features, even among affected individuals within the same family.

Common features of Type 1 Perrault syndrome:

- Hearing loss in both ears (bilateral sensorineural hearing loss, SNHL)
- In females only, ovaries that don't function as expected (ovarian dysfunction)
- Fertility in males is affected occasionally

Additional features associated with Type 2 Perrault syndrome:

- Neurological features
- Metabolic conditions

Common features of Type 1 Perrault syndrome

Hearing loss in both ears (bilateral sensorineural hearing loss (SNHL))

The degree of hearing loss and the age of onset varies and does not depend on whether the person affected is male or female. For some, hearing loss is profound and present at birth (congenital) and may be picked up at the newborn hearing screening; for others, hearing loss is moderate, with early childhood onset, and may become more severe over time. Hearing loss should be assessed as soon as noted and consideration given to interventions such a cochlear implant (CI), a surgically implanted electronic device that improves hearing. This can improve a person's ability to understand speech and hear more sounds, thereby improving a child's ability to acquire speech and language skills. Since children are at risk of speech delay, parental concerns should be acted on early and home- or school-based therapy provided.

Unique publishes separate guides to Hearing loss

In females only, ovaries that don't function as expected (ovarian dysfunction)

Ovarian dysfunction can lead to a range of conditions, from absent or underdeveloped ovaries (ovarian dysgenesis) resulting in a failure to start periods (menstruation) by the expected age, to the early loss of the normal function of the ovaries before the age of 40 resulting in early menopause (premature ovarian insufficiency, POI). The degree to which girls and women with Perrault syndrome are affected by ovarian dysfunction varies, from those who are unaffected to those with POI. A few women with Perrault syndrome resulting in POI are known to have had children before experiencing premature menopause.

Fertility in males is affected occasionally

The ability of men with Perrault syndrome to have children is usually unaffected. There are rare reports of males with conditions that reduce fertility, due to conditions such as the absence of sperm in their semen (azoospermia) and the underdevelopment of male internal and/or external genitalia and physical characteristics (undervirilization). More research in this area is needed since most of those who are affected were diagnosed before puberty, at the same time that they were diagnosed with hearing loss.

Additional features associated with Type 2 Perrault syndrome

Neurological features

A wide range of features affecting the brain, spinal cord and nerves, including learning (intellectual) disability and movement disorders affecting balance and walking (ataxia) have been described. The presence of these features appears to be associated with only some of the genes that cause Perrault syndrome (see below).

Metabolic conditions

A small number of individuals with Perrault syndrome have been described to have altered blood glucose levels in childhood, either low (hypoglycaemia) or high (diabetes). Rarely, low levels of thyroid hormone have been described (hypothyroidism).

In some babies affected by a severe form of Perrault syndrome, the condition presents as a neurometabolic condition. This can have many features, including delay in reaching developmental milestones (rolling, sitting, walking, smiling), epilepsy, and metabolic crisis (raised lactate levels in blood or cerebrospinal fluid). This is similar to severe childhood-onset mitochondrial disorders.

Features associated with different causative genes

The likelihood of an individual having certain features that are associated with Perrault syndrome varies and depends to some degree on which of the 12 genes in that individual carries the pathogenic variants. For example, Perrault syndrome caused by pathogenic variants in the *HARS2* gene is rarely associated with neurological features, while Perrault syndrome caused by pathogenic variants in the *HSD17B4* gene or *TWNK* gene is strongly associated with neurological features.

The features associated with each gene are listed below. As more people are diagnosed, and information is shared, the range of symptoms and features, and the likelihood of a child or adult having these features, will become clearer:

CLPP-related Perrault syndrome

- Thirty-one individuals reported to date [2025].
- Type 1 features:
  - Sensorineural hearing loss (SNHL), which is usually present from birth and is severe to profound.
  - Varying degrees of ovarian dysfunction, ranging from ovarian dysgenesis to premature ovarian insufficiency (POI).
  - Infertility in some males due to the absence of sperm in their semen (azoospermia).
- Type 2 features:
  - Learning difficulties.
  - Autism spectrum disorder (ASD).
  - Seizures (epilepsy), a sudden and unexpected change in the electrical activity in the brain. Depending on the parts of the brain affected, symptoms vary, but include temporary confusion, uncontrollable jerking movements, and loss of consciousness or awareness.
  - A clumsy, staggering, wide-based walking style (spastic ataxic gait).
  - Uncontrollable muscle spasms, which can be painful (dystonia).

DAP3-related Perrault syndrome

- Five individuals reported to date [2025].
- Type 1 features:
  - SNHL. Hearing loss which is usually present from birth and is profound.
  - Ovarian dysfunction. POI.

- No fertility issues in males have been reported.

- Type 2 features:
  - A varying degree of intellectual disability, ranging from mild to severe.
  - Seizures (epilepsy).
  - Inherited white matter disorders (IWMDs) affecting the white matter of the brain and spinal cord (also known as diffuse leukodystrophy). IWMDs can cause impaired mobility, vision, speech and hearing; an inability to swallow; and loss of thinking and reasoning (cognitive) skills over time.

ERAL1-related Perrault syndrome

- Four individuals reported to date [2025].
- Type 1 features:
  - SNHL. Hearing loss may be present from birth and profound, or develop in early in childhood and worsen over time with varying degrees of severity.
  - Varying degrees of ovarian dysfunction, ranging from ovarian dysgenesis to POI.
  - No fertility issues in males have been reported.
- No Type 2 features reported.

GGPS1-related Perrault syndrome

- Fourteen individuals reported to date [2025].
- Type 1 features:
  - SNHL. Hearing loss is present from birth and slowly gets worse over time
  - Varying degrees of ovarian dysfunction, ranging from ovaries that are unaffected to absent ovaries.
  - No fertility issues in males have been reported.
- Type 2 features:
  - Muscle weakness (muscular dystrophy) that begins early in life and gets worse over time. May affect muscles throughout the body (generalised muscular dystrophy) or only those closest to the torso, such as the hips and shoulders (proximal muscular dystrophy).

HARS2-related Perrault syndrome

- Twenty-four individuals reported to date [2025].
- Type 1 features:
  - SNHL. Hearing loss may be present from birth and profound but usually develops in early childhood and slowly gets worse over time with a varying degree of severity.
  - Varying degrees of ovarian dysfunction, ranging from those who are unaffected to ovarian dysgenesis.
  - No fertility issues in males have been reported.
- Type 2 features:
  - Rarely, individuals have cerebellar ataxia, a movement disorder that affects the ability to coordinate muscle movement. May affect balance and gait, limb movement, eye movement and vision (oculomotor control), as well as thinking and reasoning (cognitive) skills.

HSD17B4-related Perrault syndrome

- Twelve individuals reported to date [2025].
- Type 1 features:
  - SNHL. Hearing loss may be present from birth and profound, or it may be mild and progressively

worsen throughout childhood.

- Varying degrees of ovarian dysfunction, ranging from ovarian dysgenesis to POI.
- No fertility issues in males have been reported.

- Type 2 features:

  All affected individuals to-date have neurologic features. Reported conditions include:

  - Learning disability.
  - A gradual degeneration of the light sensitive cells of the retina of the eye causing a slow loss of vision (retinitis pigmentosa).
  - A type of cerebral palsy that mainly affects the legs, causing muscle stiffness and poor motor control (spastic diplegic cerebral palsy). This can lead to mobility challenges and delayed development.
  - Damage to the nerves located outside of the brain and spinal cord (peripheral neuropathy) that play a role in how we move (motor nerves) and respond to our senses e.g. touch, taste, smell (sensory nerves) that gets worse over time (progressive sensory and motor peripheral neuropathy).
  - Cerebellar ataxia (see HARS2-related Perrault syndrome on page 6).

LARS2-related Perrault syndrome

- Thirty-six individuals reported to date [2025].
- Type 1 features:

  - SNHL. Hearing loss may be present from birth and profound but usually develops early in childhood. Can get worse over time with a varying degree of severity of hearing loss.
  - A few individuals with no hearing impairment have been reported.
  - Varying degrees of ovarian dysfunction, ranging from ovarian dysgenesis to POI.
  - No fertility issues in males have been reported, but underdevelopment of male internal and/or external genitalia and physical characteristics has been noted (undervirilization).

- Type 2 features:

  - Progressive loss of thinking and reasoning (cognitive) skills.
  - Seizures.
  - Leukodystrophy (see DAP3-related Perrault syndrome on page 6).

MRPL49-related Perrault syndrome

- Fourteen individuals reported to date [2025].
- Type 1 features:

  - Profound bilateral hearing loss, which develops in childhood.
  - Varying degrees of ovarian dysfunction, ranging from ovarian dysgenesis to POI.
  - No fertility issues in males have been reported.

- Type 2 features:

  - Learning disability.
  - The loss of brain cells (neurons) and the connections between them (cerebral atrophy).
  - Progressive vision loss due to the gradual degeneration of the light-sensitive cells in the retina of the eye (retinal dystrophy).
  - Leukodystrophy, especially affecting a part of the brain called the globus pallidus (see DAP3-related Perrault syndrome on page 6).

PEX6-related Perrault syndrome

- One individual reported to date [2025].

- Type 1 features:
  - SNHL, which developed in childhood.
  - Ovarian dysfunction. POI.
  - No fertility issues in males have been reported.
- Type 2 features:
  - Learning disability.
  - Retinal dystrophy (see MRPL49-related Perrault syndrome on page 7).
  - Leukodystrophy (see DAP3-related Perrault syndrome on page 6).
  - Peripheral neuropathy (see HSD17B4-related Perrault syndrome on page 7).

PRORP-related Perrault syndrome

- Eleven individuals reported to date [2025].
- Type 1 features:
  - SNHL, ranging from mild to severe hearing loss.
  - Individuals with no hearing impairment have also been reported.
  - Ovarian dysfunction, ranging from POI to those who are unaffected.
  - No fertility issues in males have been reported.
- Type 2 features:
  - Learning disability.
  - Leukodystrophy (see DAP3-related Perrault syndrome on page 6).

RMND1-related Perrault syndrome

- Six individuals reported to date [2025].
- Type 1 features:
  - SNHL. Hearing loss ranging from moderate to severe.
  - Ovarian dysfunction. POI.
  - No fertility issues in males have been reported.
- Type 2 features:
  - Learning disability.
  - Leukodystrophy (see DAP3-related Perrault syndrome on page 6).

TWNK-related Perrault syndrome

- Eleven individuals reported to date [2025].
- Type 1 features:
  - SNHL. Hearing loss ranging from moderate to severe (includes problems related to the transmission of sound from the inner ear to the brain (auditory neuropathy)).
  - Ovarian dysfunction. Ranging from ovarian dysgenesis to POI.
  - No fertility issues in males have been specifically reported, but there is evidence of reduced fertility in males.
- Type 2 features:

  All affected individuals to-date have neurologic features. Reported conditions include:

  - Problems with balance and coordination (ataxia).
  - Peripheral neuropathy (see *HSD17B4*-related Perrault syndrome on page 7).

How common is Perrault syndrome?

It is difficult to say. Currently, more than 170 people with Perrault syndrome have been reported in the medical literature. Males and females are affected in equal numbers. As this is a condition due to changes inherited in a gene from each parent, Perrault syndrome is more common in families where relatives have children. The condition is under-recognised, and we are working to increase awareness.

Why did this happen?

Gene variants happen naturally and are not due to anyone's diet, environment or lifestyle. We all have our own unique set of gene variants, and most of these DNA changes have no obvious effect. This is often the case for autosomal recessive conditions where a change to one copy of the gene is not expected to cause any symptoms or features, so a parent would be unaware they had it. It is important to recognize that no one should be blamed for variants in their DNA and no parent is at fault.

In most people identified so far with Perrault syndrome, each parent passed on a copy of a gene with a pathogenic variant to their child with the condition. The affected child therefore inherited two altered copies of one of the 12 known causative genes, one from each parent. In a small number of people with Perrault syndrome the genetic change has not been identified.

Can it happen again?

The chance of having another child affected by a rare autosomal recessive gene condition depends on the genetic code of the parents. If both parents are known to unaffected carriers of a Perrault syndrome-related pathogenic variant in the same causative gene, theoretically one child in four (25 per cent) would have Perrault syndrome, two (50 per cent) would be unaffected carriers (like the parents) and one (25 per cent) would be unaffected and not a carrier. This chance resets for each pregnancy.

All the biological children of a person with Perrault syndrome will be unaffected carriers of the pathogenic variant inherited from the person with Perrault syndrome. However, most females with Perrault syndrome will not be able to have children without assisted reproduction.

Once a person has been diagnosed with Perrault syndrome, genetic testing can be carried out on relatives who may also carry the Perrault syndrome-related pathogenic variants identified in the affected person, including siblings of an affected child who should be tested as soon as possible after they are born.

Each family situation is different and there are rare cases where the inheritance of Perrault syndrome may be more complicated. A clinical geneticist or genetic counsellor should be able to give you specific advice for your family.

Family planning

Since Perrault syndrome is linked to an increased chance of fertility issues in females - as a result of ovarian dysfunction, which may be associated with conditions such as POI - and less commonly decreased fertility in males, genetic counselling should also address possible implications for all family members, including young adults who are known to be carriers or could be carriers. This could include discussions relating to different treatments that can be performed to help achieve pregnancy (assistive reproductive technology), and the potential to use prenatal and preimplantation genetic testing.

Can Perrault syndrome be cured?

At present, there is no cure or specific treatment for Perrault syndrome. However, early diagnosis means that appropriate support, monitoring and management can be put in place for individuals and their families.

Management recommendations

No clinical practice guidelines for Perrault syndrome have been published. The following recommendations have been provided by clinicians, who have personal experience of managing/treating individuals with Perrault syndrome, to improve quality of life and reduce complications.

Immediately following diagnosis

When not carried out as part of the diagnostic process, an evaluation of the features of Perrault syndrome that are present in the person who has been diagnosed should be carried out. This can determine which of the features of Perrault syndrome are present and how severe they are.

The following assessments are recommended:

- An audiological (hearing) assessment to determine the severity of hearing loss and ensure early intervention. Perrault syndrome is associated with a distinctive audiological profile, which can aid diagnosis by distinguishing it from other possible causes.
- A neurologic assessment to determine whether conditions such as ataxia, peripheral neuropathy and learning (intellectual) disability are present. Brain magnetic resonance imaging (MRI), a technique that can be used to visualise the brain, should be carried out if there are any changes to the white matter of the brain.
- Levels of hormones involved in the reproductive system, such as oestrogen and gonadotrophins (luteinising hormone (LH) and follicle-stimulating hormone (FSH), should be measured. An MRI or ultrasound examination of the ovaries should be performed.
- A physical examination to look for evidence of underdevelopment of male genitalia and physical characteristics (undervirilization) should be carried out.
- Genetic counselling for affected individuals and their families.
- Signposting to family support and resources.

Supportive care

How a person with Perrault syndrome is cared for is likely to require co-ordinated care by a multidisciplinary team of specialists, including:

Audiologists - health professionals who diagnose, treat, and help manage a hearing or balance condition.

Otorhinolaryngologists - doctors dealing with conditions of the ear, nose and throat.

Paediatricians - doctors specialising in the physical, mental and social health of children from birth to young adulthood.

Neurologists - doctors dealing with conditions of the brain, spinal cord and nervous system, including seizures. This can include some musculoskeletal conditions.

Endocrinologists - doctors dealing with hormones and their effects on the body (for example, conditions such as infertility and diabetes).

Urologists - doctors who specialise in diagnosing and treating conditions affecting the urinary system.

Geneticists - doctors dealing with diagnosis and management of genetic conditions.

Specialist nurses and/or other healthcare professionals may need to systematically and comprehensively plan an affected person's treatment.

Treatments and therapies

Treatment will depend on the specific features and symptoms experienced by the person with Perrault syndrome but may include:

Assessment and treatment by a multidisciplinary team including an audiologist and speech and language therapist to determine the best interventions, such as hearing aids and cochlear implants, and help address speech, language and communication difficulties.

Hormone treatment to induce puberty and maintain the menstrual cycle in females, and later oestrogen replacement therapy until the age of ~50 years to decrease the risk of cardiovascular disease affecting the heart and blood vessels, and weak and brittle bones (osteoporosis).

Assisted reproduction through *in vitro* fertilization (IVF), possibly using donor eggs or sperm. Egg freezing may also be recommended for women with POI when menopause happens early.

Treatments necessary for any neurologic conditions, such as pain relief.

Physiotherapy (PT) and occupational therapy (OT) to help with movement disorders.

Surveillance

- It is recommended that the following evaluations are carried out to monitor an individual's existing Perrault syndrome symptoms, how they respond to care and treatment, and whether any new symptoms emerge over time:
- Annual audiologic assessments to monitor and assess possible progressive hearing impairment.
- Regular meetings with an endocrinologist for assessment of any side effects and responses to hormone therapy.
- Assessment of the need for social support, such as respite care and home nursing care, or genetic counselling and family planning advice.
- Those with hearing loss should avoid oxotoxic medications (substances that are toxic to the ear), such as aminoglycosides, if alternative medications are available. They should also avoid exposure to loud noise, which can contribute to progressive hearing loss.

Is there any research into new treatments for Perrault syndrome?

The research group of Professor Bill Newman at the University of Manchester is undertaking work which will hopefully lead to potential treatments. Other research groups across the world have expertise in mitochondrial disorders and will be adding to this endeavour.

Details of clinical trials related to a particular condition or gene can be found at **ClinicalTrials.gov** and **EU Clinical Trials Register**.

Sources

The information in this booklet is drawn from the published medical literature and information from Unique members. In 2025, Unique had 11 members with ZTK syndrome. The first-named author and publication date for articles in the medical literature are given to allow you to look for the abstracts or original articles on the internet in PubMed (**pubmed.ncbi.nlm.nih.gov/**).

References

Li et al. (2014) [updated 2025] Perrault Syndrome Overview. GeneReviews® [Internet] Seattle (WA) * **Link to article**

National Organisation for Rare Disorders (NORD) - rarediseases.org/rare-diseases/perrault-syndrome/ * **Link to article**

Genetics and Rare Diseases Information Center (GARD) - rarediseases.info.nih.gov/diseases/2542/perrault-syndrome * **Link to article**

Note: an asterisk indicates articles which are "open access" and available to everyone at **pubmed.ncbi.nlm.nih.gov**

Websites, Facebook groups and Support Groups

National deaf children's society (NDCS) - Dedicated to creating a world without barriers for deaf children and young people.

Royal National Institute for Deaf People (RNID) - National charity supporting people in the UK who are deaf, have hearing loss or tinnitus.

Alexander Graham Bell Association for the Deaf and Hard of Hearing - Works globally to empower individuals who are deaf or hard-of-hearing, providing support for them to listen, speak, and thrive.

American Society for Deaf Children - Supporting parents of children who are deaf and hard-of-hearing.

National Association of the Deaf (NAD) - A civil rights organisation for deaf and hard-of-hearing individuals.

Fertility Network UK - Supports fertility clinics to help them to support, and show their commitment to, the emotional wellbeing of their patients.

RESOLVE: The National Infertility Association - Provides free support in communities around the UK for people struggling to build their families.

The InterNational Council on Infertility Information Dissemination (INCIID) – A nonprofit organisation that helps individuals and couples explore their family-building options.

Alex TLC - Provides support and information to people affected by leukodystrophy.

Ataxia UK - The leading national charity in the UK for people affected by any type of ataxia.

CureARS - Spreading awareness, connection and support to affected families and funding research for the ultra-rare mitochondrial ARS.

Metabolic Support UK (MDUK) - Organisation for Inherited Metabolic Disorders (IMDs), supporting, building communities, and advocating for those living with IMDs.

The Lily Foundation - Improving the lives of patients and families affected by mitochondrial disease.

Inform Network Support

Rare Chromosome Disorder Support Group
The Stables, Station Road West, Oxted, Surrey, RH8, 9EE, UK
Tel: +44(0)1883 723356
help@rarechromo.org I rarechromo.org

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This information 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.

This guide was compiled by Unique (CA) and verified by Professor Bill Newman (PhD FRCP FMedSci), Consultant in Clinical Genetics, Manchester University NHS Foundation Trust (MFT) and University of Manchester, UK.

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