CONDITION

Rapp Hodgkin syndrome

Rapp Hodgkin syndrome is an inherited condition affecting the hair, teeth, nails, skin, sweating, and facial development.

WHAT IT IS
Inherited ectodermal development condition in the TP63 spectrum

BODY SYSTEM
Skin, hair, teeth, nails, eyes, and facial development

MAIN SYMPTOMS
Sparse hair, abnormal teeth, reduced sweating, cleft lip or palate

WHAT CAUSES IT
Inherited or new TP63 gene change

HOW IT IS DIAGNOSED
Clinical assessment and TP63 genetic testing

MAIN TREATMENT
Coordinated skin, dental, eye, cleft, and heat care

CAN IT BE CURED?
No - the genetic change is lifelong

IS IT AN EMERGENCY?
Sometimes - severe overheating or breathing difficulty

LONG-TERM OUTLOOK
Lifelong features, often manageable with coordinated care

Overview

Rapp Hodgkin syndrome is a rare genetic condition that affects structures formed from the ectoderm. The ectoderm is an early layer of a developing embryo that helps form the skin, hair, nails, teeth, sweat glands, and parts of the eyes and face.

The condition is a type of ectodermal dysplasia. "Dysplasia" means that a body structure developed differently. Common features include sparse hair, missing or unusually shaped teeth, abnormal nails, reduced sweating, and a cleft lip or palate.

Rapp Hodgkin syndrome is caused by a change in the TP63 gene. It overlaps closely with ankyloblepharon-ectodermal defects-cleft lip/palate syndrome, usually shortened to AEC syndrome or called Hay-Wells syndrome. Specialists now generally consider Rapp Hodgkin syndrome part of the same TP63-related spectrum. The older name is still used, especially

when a person's features match the traditional Rapp Hodgkin pattern.

The condition does not affect everyone in the same way. Some people mainly need dental and heat-management care. Others need treatment for a cleft palate, skin problems, blocked tear ducts, hearing concerns, or feeding and speech difficulties.

Symptoms

Features are usually present from birth or become clearer during childhood. A person may have only some of the following:

Some people within the overlapping AEC spectrum have fragile skin or raw areas where the top layer of skin is missing. These erosions may be present on the scalp or other parts of the body. They can heal slowly and may become infected.

A cleft palate can make it difficult for a baby to create suction during feeding. Later, it may affect speech or allow fluid to enter the middle ear. Missing or abnormal teeth can affect chewing, speech, jaw development, and appearance.

Reduced sweating is medically important because sweating helps cool the body. A person may become too hot during warm weather, exercise, fever, or time in a hot room. How much heat intolerance occurs is not always clear from appearance alone.

Intelligence is not usually affected directly by Rapp Hodgkin syndrome. Hearing, speech, discomfort, repeated procedures, or missed school can still influence learning and daily life.

- Thin, sparse, coarse, or slow-growing scalp hair
- Sparse or absent eyebrows and eyelashes
- Missing teeth, widely spaced teeth, or small cone-shaped teeth
- Thick, ridged, unusually shaped, or poorly developed nails
- Reduced sweating and difficulty coping with heat
- Dry or sensitive skin
- A cleft lip, cleft palate, or both
- A narrow nose, small mouth, or other characteristic facial features
- Blocked or poorly developed tear ducts
- Dry, irritated, or watery eyes
- Repeated ear problems or hearing loss

Causes and risk factors

Rapp Hodgkin syndrome is caused by a disease-causing change, also called a pathogenic variant, in the TP63 gene. This gene gives cells instructions for making the p63 protein, which is important during the development of the skin, limbs, face, teeth, and other ectodermal structures.

The condition follows an autosomal dominant inheritance pattern. This means one altered copy of the gene is enough to cause the condition. If a parent has the genetic change, each child has a 1 in 2 chance of inheriting it. The features and their severity cannot be predicted from that chance.

Sometimes the genetic change occurs for the first time in an egg, sperm, or early embryo. This is called a new, or de novo, variant. In that situation, neither parent necessarily has the condition.

People of any sex can be affected. A family history raises the chance of having the condition, but the absence of a known family history does not rule it out.

Diagnosis

Diagnosis begins with a medical and family history and an examination of the hair, teeth, nails, skin, sweating, eyes, mouth, and face. A clinician may also check hearing, tear drainage, feeding, speech, and growth.

A genetic test that finds a disease-causing TP63 variant can confirm the diagnosis. Testing may use a panel that examines TP63 together with other genes linked to ectodermal dysplasia. A clinical genetics service can help explain what a result means for the person tested and their relatives.

Genetic results need careful interpretation. Some gene changes have an uncertain meaning, and a negative test may not fully explain clear ectodermal features. The diagnosis may therefore combine genetic findings with the person's physical features.

Additional assessments are chosen according to symptoms. They may include dental X-rays to identify missing teeth, hearing tests, an eye examination, or evaluation by a cleft lip and palate team.

Treatment and management

There is no treatment that removes the underlying TP63 genetic change. Care focuses on protecting affected tissues, supporting development, and treating specific problems.

A coordinated plan may involve dermatology, dentistry, orthodontics, clinical genetics, ophthalmology, audiology, ear, nose and throat care, and a cleft lip and palate team.

Skin care may include gentle washing, moisturisers, suitable dressings for erosions, and treatment of confirmed infections. Persistent scalp erosions often need specialist wound care because repeated trauma can delay healing.

Heat management is important when sweating is reduced. Helpful measures can include access to water, shade, air conditioning or fans, light clothing, cooling breaks, and adjusting activity in hot weather. Schools and workplaces may need a written plan that allows the person to cool down promptly.

Dental care often starts early. Depending on age and tooth development, options may include protective dental care, dentures, crowns, orthodontic treatment, or implants later in life. Treatment plans are usually adjusted as the jaws and remaining teeth grow.

A cleft lip or palate may require surgery and support with feeding, hearing, dental development, and speech. Babies who cannot feed effectively may need specialised bottles or feeding advice from their cleft team.

Blocked tear ducts, dry eyes, eyelid differences, or repeated eye irritation should be assessed by an eye specialist. Management may include eye-surface protection or procedures to improve tear drainage.

Hearing tests are useful when there are repeated ear infections, a cleft palate, delayed speech, or concern about hearing. Hearing aids or other support may be offered when needed.

Hair changes do not always require medical treatment. Some people choose wigs, hairpieces, cosmetic eyebrow products, or other appearance-related support. These are personal choices rather than medical requirements.

Complications

Overheating is one of the main preventable complications. Without enough sweating, the body may struggle to release heat. Severe overheating can lead to dehydration, confusion, fainting, or seizures.

Open skin erosions can be painful and may become infected. Healing can cause scarring or areas where hair does not regrow.

Dental differences may make chewing difficult and can affect nutrition, speech, and

confidence. Remaining teeth may be exposed to extra wear, so preventive dental care is especially important.

Poor tear drainage or a dry eye surface can cause repeated irritation and infection. Untreated eye-surface damage may threaten vision, although this is not inevitable.

Cleft palate and middle-ear problems can contribute to hearing loss and unclear speech. Visible differences, repeated medical care, or unwanted attention may also affect emotional wellbeing. Psychological or peer support can be included as part of care.

Prevention

Rapp Hodgkin syndrome cannot be prevented through diet, lifestyle, or pregnancy behaviour. It results from a genetic change that is inherited or occurs for the first time.

Some complications can be reduced through heat precautions, early dental care, skin protection, hearing checks, eye care, and treatment of feeding or cleft-related problems.

If a specific TP63 variant is known in a family, genetic counselling can explain reproductive options. These may include testing during pregnancy or testing embryos created through in vitro fertilisation. Whether to use these options is a personal decision.

Living with it and what to expect

Rapp Hodgkin syndrome is lifelong, but it is not usually a steadily worsening condition. Hair, tooth, nail, and sweating differences generally continue into adulthood. Dental needs change as the jaws grow and teeth develop, so treatment may take place in stages.

Skin erosions within the AEC spectrum may improve with age, although some people continue to have fragile or inflamed areas. Heat intolerance may remain even when a person learns how to manage it well.

Children may have periods of more intensive care for feeding, cleft repair, hearing, speech, or dental development. In adulthood, care often shifts toward maintaining dental work, protecting the eyes and skin, and managing heat exposure.

Many people attend mainstream school, work, exercise with suitable precautions, and have families. Practical adjustments may be needed for hot classrooms, outdoor work, sports, dental appointments, or hearing and speech support. The amount of support required is individual and may change over time.

Routine follow-up should focus on the problems a person actually has. Regular dental care is

often needed, while eye, hearing, skin, or cleft follow-up may be arranged at different intervals.

When to get medical help

Call emergency services now if someone has trouble breathing, becomes confused or unresponsive, has a seizure, or collapses. These symptoms may occur with severe overheating or another serious problem.

Move a person with possible overheating to a cooler place while arranging help. Remove unnecessary outer clothing and begin safe cooling if possible. Do not delay emergency care when there is confusion, fainting, or a seizure.

Get urgent medical advice for:

Arrange routine follow-up for new dental pain, changes in hearing, repeated ear or eye infections, worsening skin problems, speech concerns, or difficulty managing heat. These usually need assessment but are not automatically emergencies.

- Eye pain, strong light sensitivity, or a sudden change in vision
- Spreading redness, swelling, pus, or fever with a skin wound
- Inability to drink or keep fluids down
- A baby who is struggling to feed, choking, or not gaining weight
- Signs of overheating that do not improve quickly with cooling

Common questions

Is Rapp Hodgkin syndrome the same as AEC syndrome?

They are now generally considered part of the same spectrum of TP63-related conditions. Rapp Hodgkin syndrome was originally described as a separate condition because its pattern of features appeared different. Genetic findings later showed substantial overlap with AEC, or Hay-Wells, syndrome.

Can Rapp Hodgkin syndrome be cured?

No current treatment removes the TP63 genetic change. Many individual effects can be treated or managed, including cleft palate, missing teeth, skin wounds, eye problems, hearing loss, and difficulty coping with heat.

Will someone with this condition always overheat?

Not everyone has the same degree of reduced sweating. Some people tolerate ordinary

activity but struggle in hot weather or during exercise. Others need more consistent cooling measures. A personalised heat plan can help identify safe activities and early warning signs.

Does Rapp Hodgkin syndrome affect intelligence?

The condition does not usually affect intelligence directly. Hearing loss, speech difficulties, discomfort, hospital appointments, or interrupted schooling may create learning barriers that need support.

Can Rapp Hodgkin syndrome be passed to children?

Yes. A person with a disease-causing TP63 variant has a 1 in 2 chance of passing it on in each pregnancy. A child who inherits the variant may not have exactly the same features or severity as their parent.

Will missing teeth or sparse hair grow normally later?

Missing permanent teeth will not appear later if the tooth buds never developed. Dental replacement can improve chewing and appearance. Sparse hair may remain thin throughout life, although its texture and coverage can change with age.

Sources

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