CONDITION

Alobar holoprosencephaly

Learn how alobar holoprosencephaly affects brain development, how it is diagnosed, and how supportive care can address feeding, seizures, breathing, and comfort.

WHAT IT IS
Severe disorder of early brain development

BODY SYSTEM
Nervous system

MAIN SYMPTOMS
Profound developmental delay, seizures, feeding difficulty, breathing problems

WHAT CAUSES IT
Genetic or chromosome changes, sometimes unknown

HOW IT IS DIAGNOSED
Prenatal ultrasound or fetal MRI, postnatal brain MRI

MAIN TREATMENT
Supportive and palliative care

CAN IT BE CURED?
No, brain formation cannot be reversed

IS IT AN EMERGENCY?
Sometimes, severe or persistent breathing trouble

LONG-TERM OUTLOOK
Profound needs with often shortened survival

Overview

Alobar holoprosencephaly is a serious condition that begins early in pregnancy as the brain is forming. Normally, the front part of the developing brain divides into right and left halves. In alobar holoprosencephaly, this separation does not occur.

"Alobar" describes the brain's structure. It is generally considered the most severe form of holoprosencephaly. The cerebral hemispheres are largely joined, and there is usually one large fluid-filled brain cavity instead of two side ventricles. Other deep brain structures may also be joined or formed differently.

Because the brain guides movement, feeding, breathing, hormone control, temperature, and development, the condition can affect many body systems. It is often found during pregnancy. Some affected pregnancies end in miscarriage or stillbirth. Babies born alive usually have

profound developmental and medical needs, although the exact course varies.

Differences involving the middle of the face may occur, such as a cleft lip or palate, closely spaced eyes, or an unusually small nose. However, facial appearance does not always show the full severity of the brain differences.

There is no treatment that can separate the brain after it has formed. Care focuses on comfort, nutrition, breathing, seizure control, hormone problems, and family goals. Families may choose different levels of medical support after discussions with maternal-fetal medicine, newborn, neurology, genetics, and palliative care teams.

Symptoms

Symptoms depend on the child's brain anatomy and associated genetic or organ differences. Some findings are visible on prenatal imaging. Others become clear after birth.

Common or important features can include:

The hypothalamus and pituitary gland may be affected. These brain structures help regulate hormones and basic body functions. A child may develop diabetes insipidus, a condition that causes unusually large amounts of dilute urine and can lead to dehydration or abnormal blood sodium. Other pituitary hormone problems are possible.

Not every child has every symptom. The brain scan is important, but it cannot predict every detail of a child's comfort, abilities, or lifespan.

- Severe delay in movement, communication, and learning
- A small head, although fluid buildup can sometimes enlarge the head
- Poor muscle tone, stiffness, abnormal movements, or a mixture of these
- Seizures
- Difficulty coordinating sucking and swallowing
- Choking, coughing, or breathing changes during feeding
- Slow growth or difficulty gaining weight
- Episodes of abnormal breathing or low oxygen
- Problems controlling body temperature, sleep, thirst, or fluid balance
- Vision or hearing impairment
- Cleft lip, cleft palate, or other differences in facial development

Causes and risk factors

Alobar holoprosencephaly results from a disruption in very early brain development. The disruption often occurs before a person knows they are pregnant. In many families, nothing a parent did caused the condition.

Chromosome differences are an important cause. Chromosomes are packages of genetic material. Holoprosencephaly may occur with trisomy 13, in which there is an extra copy of chromosome 13, or with other missing, extra, or rearranged chromosome material. These conditions may also affect the heart, kidneys, limbs, and other organs.

Changes in individual genes can also interfere with the signals that divide the developing brain. Genes associated with holoprosencephaly include SHH , ZIC2 , SIX3 , and TGIF1 , among others. A gene change may be new in the child, or it may be inherited from a parent. Some inherited changes have variable effects, meaning relatives with the same change may have mild findings, severe findings, or no obvious symptoms.

Pregestational diabetes, meaning diabetes that was present before pregnancy, is a recognized risk factor. A risk factor increases the chance of a condition but does not prove what caused it in an individual pregnancy. Researchers have also studied certain exposures during early pregnancy, but a specific exposure cannot be identified in many cases.

The chance of recurrence in another pregnancy depends on the cause. It may be low when the event was isolated and new, or substantially higher with certain inherited gene or chromosome changes. A genetic counselor can explain recurrence using the family's actual test results rather than a general estimate.

Diagnosis

Alobar holoprosencephaly may be suspected on a prenatal ultrasound. A detailed ultrasound, sometimes called targeted neurosonography, examines the brain, face, heart, growth, and other organs. Fetal MRI can provide additional pictures of the brain, especially later in pregnancy, but it does not replace a full ultrasound assessment.

Prenatal imaging usually shows a brain that has not separated into two hemispheres, a single large central fluid cavity, and joined deep brain structures. Imaging can identify the anatomical form, but it may not identify the underlying genetic cause.

Parents may be offered diagnostic genetic testing through chorionic villus sampling or amniocentesis. Depending on the situation, testing may include:

Each test has limits. A negative genetic result does not rule out holoprosencephaly or

guarantee that the condition will not recur.

After birth, diagnosis is confirmed with examination and brain imaging, usually MRI. Ultrasound or CT may be used when MRI is not immediately practical. Clinicians may also check blood sugar, sodium, urine output, pituitary hormones, hearing, vision, swallowing, and heart or kidney structure.

A clinical geneticist and genetic counselor can review the child's features, test results, and family history. Testing the biological parents may be needed to understand whether a genetic finding was inherited.

- Chromosome analysis to look for large chromosome changes
- Chromosomal microarray to identify smaller missing or extra sections of DNA
- A gene panel focused on holoprosencephaly-related genes
- Exome or genome sequencing when earlier testing does not provide an answer

Treatment and management

No treatment can reverse the underlying brain formation. Management is individualized and may include life-prolonging treatment, comfort-focused treatment, or a combination. Perinatal or pediatric palliative care can support symptom control and family decision-making alongside other medical treatment. It is not limited to end-of-life care.

Before delivery, families can meet with maternal-fetal medicine, neonatology, genetics, and palliative care specialists. A birth plan may document preferences about monitoring, breathing support, feeding, resuscitation, religious or cultural practices, and time with the baby. Plans can be changed as the child's needs become clearer.

Feeding requires careful assessment. Some children can take food by mouth with modified positioning, pacing, or food texture. Others cannot swallow safely. A feeding specialist may perform a swallowing evaluation. Tube feeding through the nose or directly into the stomach may be considered when it supports the family's goals. Tube feeding can provide nutrition but does not eliminate every risk of reflux or aspiration, which means food or saliva entering the airway.

Seizures are treated with antiseizure medicine selected according to the seizure type, other health problems, and treatment goals. Families may receive a written seizure plan describing what to observe and when emergency medicine or emergency services are needed.

Hormone and fluid problems require laboratory monitoring. Diabetes insipidus and other pituitary disorders may need specialist care and medication. Treatment must be adjusted using

urine output, growth, symptoms, and blood tests because both dehydration and excessive fluid retention can be harmful.

Other supportive care may include oxygen or breathing assistance, management of saliva and airway secretions, physical therapy for positioning and comfort, and treatment of muscle stiffness. Hearing and vision services may help a child respond to their surroundings.

Cleft lip or palate surgery, treatment of fluid buildup around the brain, or other procedures may be discussed. The expected benefit, burden of recovery, anesthesia risk, and family's goals should be considered before an operation.

Comfort care remains important with any treatment plan. This can include relief of pain, breathlessness, agitation, dry mouth, and feeding-related distress. Social workers, chaplains, bereavement professionals, and community nursing services can support the whole family.

Complications

Children with alobar holoprosencephaly are medically fragile. Serious complications can include aspiration, pneumonia, repeated seizures, breathing failure, dehydration, abnormal sodium levels, and poor nutrition. Reduced mobility can contribute to joint stiffness, skin pressure injuries, and discomfort.

Pituitary or hypothalamic dysfunction can disrupt temperature, sleep, thirst, and hormone control. Infections or feeding problems may become harder to manage when breathing and neurological function are already limited.

Some babies die before or soon after birth. Others survive longer with extensive support. It is usually not possible to predict an exact lifespan from the diagnosis alone. The presence of chromosome or organ abnormalities, breathing problems, safe feeding ability, and the family's chosen level of medical intervention can all affect the course.

Follow up

Follow-up should be coordinated around the child's needs and the family's goals. Visits may involve primary care, neurology, endocrinology, gastroenterology, nutrition, pulmonology, rehabilitation, genetics, and palliative care. When possible, combining appointments or using home-based services can reduce the burden of travel.

Routine follow-up may monitor growth, feeding tolerance, seizures, breathing, muscle tone, skin health, urine output, sodium, and other hormone measures. Equipment, medicines, and the emergency care plan should be reviewed regularly. Families should also be offered practical

and emotional support, including respite care when available.

Contact the child's clinical team promptly for a new feeding cough, repeated vomiting, worsening constipation, changes in seizure pattern, reduced urine, unusually high urine output, fever, new discomfort, or trouble using medical equipment.

Seek urgent or emergency care for:

For children receiving comfort-focused care at home, the plan may direct families to call the palliative care or hospice team instead of emergency services. Families should know whom to contact at any hour.

- Severe or persistent trouble breathing, blue or gray lips, or long pauses in breathing
- A seizure lasting longer than the child's emergency plan allows, or repeated seizures without recovery
- Choking with ongoing breathing difficulty
- Extreme sleepiness, poor responsiveness, or a sudden major change from the child's usual behavior
- Signs of severe dehydration, such as very little urine with marked drowsiness
- Any situation that the child's written emergency plan identifies as an emergency

Sources

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