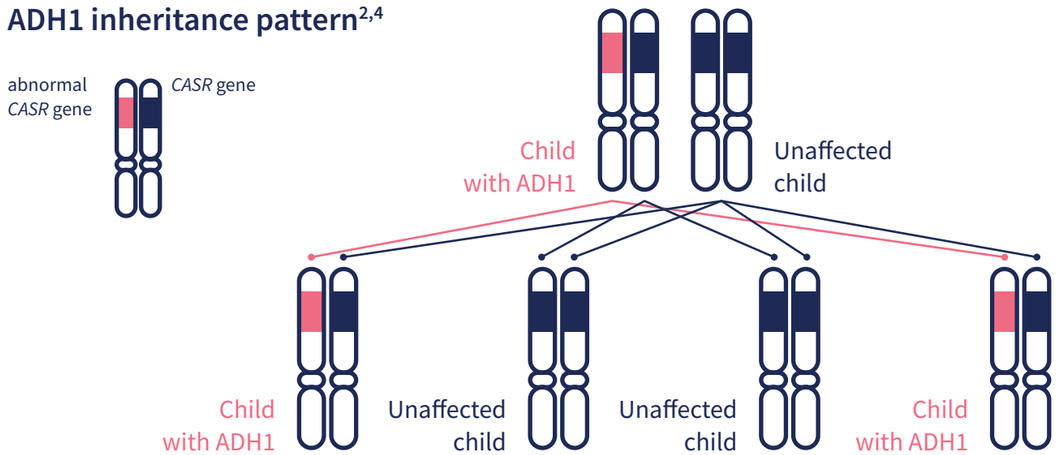
Who gets or passes down ADH1?

ADH1 is inherited, or passed from one generation to the next, in an autosomal dominant pattern.[2,4] In an autosomal dominant inheritance pattern, one copy of the *CASR* gene with an abnormal change is enough to cause the condition.[2] In most cases, a person with ADH1 gets the abnormal *CASR* gene from one parent with ADH1.[2] Each child of someone with ADH1 has a 50% chance of inheriting the abnormal gene for ADH1.[9] Some people do not inherit ADH1. Instead, ADH1 can be caused by a spontaneous change in the *CASR* gene. Thus, people can develop ADH1 with no history of the condition in their family.[2] If someone is diagnosed with ADH1, their parents also should be tested for the abnormal *CASR* gene.[10]

ADH1 inheritance pattern[2,4]



Whether you're seeking more information, navigating diagnostic uncertainty, or wish to share a patient story, we invite you to get in touch.

References: 1. Roszko et al. J Bone Miner Res. 2022;37:1926–1935. **2.** MedlinePlus. https://medlineplus.gov/genetics/condition/autosomal-dominanthypocalcemia/. Accessed July 24, 2023. **3.** Cinque et al. J Clin Endocrinol Metab. 2017;102:3961–3969. **4.** Mannstadt et al. Poster presented at: ENDO 2023; June 15-18, 2023; Chicago, IL, USA. **5.** Mannstadt et al. J Bone Miner Res. 2022;37:2615–2629. **6.** Rejnmark et al. Endocrinol Metab (Seoul). 2015;30:436–442. **7.** MedlinePlus. https://medlineplus.gov/genetics/gene/casr. Accessed July 24, 2023. **8.** Johns Hopkins Medicine. https://www.hopkinsmedicine.org/research/understanding-clinical-trials/clinical-research-what-is-it.html. Accessed July 24, 2023. **9.** National Human Genome Research Institute. https://www.genome.gov/genetics-glossary/Autosomal-Dominant-Disorder. Updated March 4, 2026. Accessed March 4, 2026. **10.** MedlinePlus. https://medlineplus.gov/ency/article/002049.htm. Accessed March 4, 2026.

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MAT-EU-ECLTX-0022

About **ADH1**

What is ADH1?

ADH1, or **A**utosomal **D**ominant **H**ypocalcemia Type **1**, is a form of hypoparathyroidism. It is an **inherited, lifelong condition**; can be found in children and adults; and is rare, affecting approximately 1 in 25,000 individuals.[1]

What are other names for ADH1?

- Autosomal dominant hypocalcemia (ADH)[2,3]
- Autosomal dominant hypoparathyroidism[2]
- Familial hypercalciuric hypocalcemia[2]
- Familial hypocalcemia[2]
- Familial isolated hypoparathyroidism[3]
- Idiopathic hypoparathyroidism[4]

People with ADH1:[2]

- Will usually have low levels of calcium in the blood, known as hypocalcemia
- Have low levels of a hormone called parathyroid hormone, which helps control the amount of calcium in the blood
- Can have high levels of calcium in their urine, known as hypercalciuria, which can lead to calcium building up in the kidneys or the formation of kidney stones
- May also have the wrong amount of other important minerals in their body, such as too much phosphate or too little magnesium

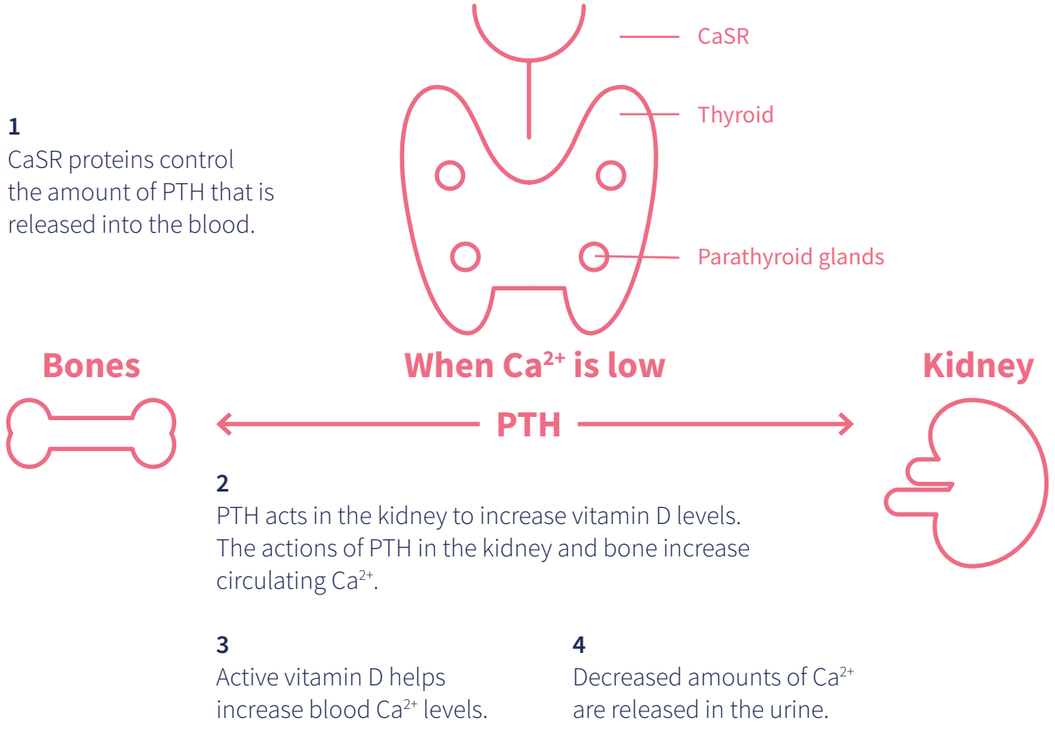
What causes ADH1?

ADH1 is caused by an abnormal change in the Calcium-Sensing Receptor (*CASR*) gene. The proteins made from this gene normally work together to detect and control the amount of calcium in the blood. In patients with ADH1, the proteins made from the abnormal *CASR* gene are more sensitive to calcium and do not work as well to control calcium levels, even when levels in the blood are low.[2] There are other abnormal genes that can cause forms of hypoparathyroidism similar to ADH1, but changes in the *CASR* gene are one of the most common.[5]

What are the symptoms of ADH1?

The symptoms of ADH1 may be **different from person to person** and may affect children and adults differently. Symptoms can appear at any age and may change over time.[4,6] One of the most common symptoms of ADH1 is sudden, strong muscle tightening, known as **muscle spasms**, in the hands and feet.[2,4] Sensations of **muscle cramping; prickling or tingling**, known as paresthesia; or twitching of the nerves and muscles in various parts of the body are also common.[2,4] People with serious ADH1 may develop short periods of uncontrolled movements, known as **seizures**, usually in infancy or childhood.[2,4]

How is CaSR involved in controlling calcium levels?



1
CaSR proteins control the amount of PTH that is released into the blood.

2
PTH acts in the kidney to increase vitamin D levels. The actions of PTH in the kidney and bone increase circulating Ca^{2+}.

3
Active vitamin D helps increase blood Ca^{2+} levels.

4
Decreased amounts of Ca^{2+} are released in the urine.

Ca^{2+}, calcium ion; **CaSR,** calcium-sensing receptor; **PTH,** parathyroid hormone.

Most common symptoms of ADH1

Brief or sudden symptoms of ADH1:[4,7,8]

Symptoms affecting the brain and muscles:
- Seizures
- Intermittent muscular spasms, known as tetany
- Paresthesia
- Muscle cramps/spasms
- Pain in bones or where bones connect, known as joints
- Feeling tired all over, known as fatigue

Symptoms affecting a person's thoughts:
- Fear or worry, known as anxiety
- Problems with thinking, remembering, or concentrating, known as brain fog

Long-term potential consequences of ADH1:[2,4,7,8]

Affecting the kidneys:
- Kidney stones
- Too much calcium in the kidneys
- Damaged kidneys that can't properly filter blood, known as chronic kidney disease

Affecting other parts of the body:
- A buildup of calcium on important regions of the brain
- Breaks in bones, known as bone fractures
- Feeling sad or hopeless and having little interest in activities, known as depression
- Heart beating too fast or too slow, known as arrhythmia