

Jervell and Lange-Nielsen Syndrome

Subjects: Genetics & Heredity

Contributor: Camila Xu

Jervell and Lange-Nielsen syndrome is a condition that causes profound hearing loss from birth and a disruption of the heart's normal rhythm (arrhythmia).

Keywords: genetic conditions

1. Introduction

This disorder is a form of long QT syndrome, which is a heart condition that causes the heart (cardiac) muscle to take longer than usual to recharge between beats. Beginning in early childhood, the irregular heartbeats increase the risk of fainting (syncope) and sudden death.

2. Frequency

Jervell and Lange-Nielsen syndrome is uncommon; it affects an estimated 1.6 to 6 per 1 million people worldwide. This condition has a higher prevalence in Denmark, Sweden, and Norway, where it affects at least 1 in 200,000 people.

3. Causes

Jervell and Lange-Nielsen syndrome is caused by mutations in the *KCNE1* and *KCNQ1* genes. These genes provide instructions for making proteins that work together to form a channel across cell membranes. These channels transport positively charged potassium atoms (ions) out of cells. The movement of potassium ions through these channels is critical for maintaining the normal functions of inner ear structures and cardiac muscle.

About 90 percent of cases of Jervell and Lange-Nielsen syndrome are caused by mutations in the *KCNQ1* gene; *KCNE1* mutations are responsible for the remaining cases. Mutations in these genes alter the usual structure and function of potassium channels or prevent the assembly of normal channels. These changes disrupt the flow of potassium ions in the inner ear and in cardiac muscle, leading to hearing loss and an irregular heart rhythm characteristic of Jervell and Lange-Nielsen syndrome.

3.1. The genes associated with Jervell and Lange-Nielsen syndrome

- *KCNE1*
- *KCNQ1*

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. Most often, the parents of a child with an autosomal recessive disorder are not affected but are carriers of one copy of the mutated gene. Some carriers of a *KCNQ1* or *KCNE1* mutation have a long QT interval with related heart abnormalities, but their hearing is normal.

5. Other Names for This Condition

- autosomal recessive long QT syndrome (LQTS)
- cardio-auditory-syncope syndrome
- cardioauditory syndrome of Jervell and Lange-Nielsen

- deafness, congenital, and functional heart disease
- Jervell-Lange Nielsen syndrome
- JLNS
- prolonged QT interval in EKG and sudden death
- surdo-cardiac syndrome

References

1. Mizusawa Y, Horie M, Wilde AA. Genetic and clinical advances in congenital long QT syndrome. *Circ J*. 2014;78(12):2827-33.
 2. Modell SM, Lehmann MH. The long QT syndrome family of cardiac ion channelopathies: a HuGE review. *Genet Med*. 2006 Mar;8(3):143-55. Review.
 3. Nakano Y, Shimizu W. Genetics of long-QT syndrome. *J Hum Genet*. 2016 Jan;61(1):51-5. doi: 10.1038/jhg.2015.74.
 4. Priori SG, Wilde AA, Horie M, Cho Y, Behr ER, Berul C, Blom N, Brugada J, Chiang CE, Huikuri H, Kannankeril P, Krahn A, Leenhardt A, Moss A, Schwartz PJ, Shimizu W, Tomaselli G, Tracy C. HRS/EHRA/APHRS expert consensus statement on the diagnosis and management of patients with inherited primary arrhythmia syndromes: document endorsed by HRS, EHRA, and APHRS in May 2013 and by ACCF, AHA, PACES, and AEPC in June 2013. *Heart Rhythm*. 2013 Dec;10(12):1932-63. doi:10.1016/j.hrthm.2013.05.014.
 5. Schwartz PJ, Crotti L. QTc behavior during exercise and genetic testing for the long-QT syndrome. *Circulation*. 2011 Nov 15;124(20):2181-4. doi:10.1161/CIRCULATIONAHA.111.062182.
 6. Schwartz PJ, Spazzolini C, Crotti L, Bathen J, Amlie JP, Timothy K, Shkolnikova M, Berul CI, Bitner-Glindzicz M, Toivonen L, Horie M, Schulze-Bahr E, Denjoy I. The Jervell and Lange-Nielsen syndrome: natural history, molecular basis, and clinical outcome. *Circulation*. 2006 Feb 14;113(6):783-90.
 7. Tranebjærg L, Samson RA, Green GE. Jervell and Lange-Nielsen Syndrome. 2002 Jul 29 [updated 2017 Aug 17]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. *GeneReviews®* [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1405/>
 8. Wang Z, Li H, Moss AJ, Robinson J, Zareba W, Knilans T, Bowles NE, Towbin JA. Compound heterozygous mutations in KvLQT1 cause Jervell and Lange-Nielsen syndrome. *Mol Genet Metab*. 2002 Apr;75(4):308-16.
-

Retrieved from <https://encyclopedia.pub/entry/history/show/11623>