

Jervell and Lange–Nielsen Syndrome

Also known as: cardioauditory syndrome, surdocardiac syndrome

Clinical Characteristics

Jervell and Lange-Nielsen syndrome (JLNS) is characterized by congenital hearing loss and a form of long Q-T syndrome. Long Q-T syndrome, in which the heart takes longer than usual to recharge between beats, results in symptoms such as tachyarrhythmia, ventricular fibrillation, and syncope (fainting). Young children with JLNS are especially prone to syncopal episodes during stress, exercise, or fright.

Jervell and Lange-Nielsen Syndrome and Hearing Loss

Individuals with JLNS typically are diagnosed with congenital, bilateral, and severe to profound sensorineural hearing loss.

Natural History

JLNS is diagnosed in both males and females, and in all ethnicities. The prevalence of the condition is estimated to be 1 to 6 per million, though some “false positives” are likely included in that number. About 0.25% of profound congenital hearing loss has been attributed to JLNS.

Hearing loss is present at birth. Long Q-T syndrome is also present, though is usually not recognized until the child is older. The prognosis for individuals with JLNS is not well-established. It is known that there is an increased risk of death in infancy, and 50% of untreated patients die before the age of 15 years. Sudden death occurs in over 25% of cases. With careful surveillance, treatment, and prevention of syncope and arrhythmia, though, affected children can live well into adulthood. The prognosis is not as good for children who have had a cardiac episode before the age of 5. Males appear to have a higher risk for cardiac arrest and sudden death than females. Intelligence is normal.

Genetics

Two genes are known to be associated with JLNS: *KCNQ1* on chromosome 11 (90% of cases), and *KCNE1* on chromosome 21. Both of these genes code for potassium channels found in the cochlea and the heart. The mutation detection rate in affected individuals is unknown. JLNS is **autosomal recessive**. This means that both parents of an affected individual must be carriers for the condition. Usually, carriers of recessive disorders are asymptomatic. However, some heterozygous carriers of JLNS have demonstrated symptoms of long-QT syndrome. As such, parents of affected children should be offered comprehensive electrocardiograph testing. These parents usually

have normal hearing. With each pregnancy, two carrier parents of JLNS have a 1 in 4 (25%) chance of having an affected child, a 2 in 4 (50%) chance of having a child who is a carrier like themselves, and a 1 in 4 (25%) chance of having a child who is neither affected nor a carrier. Overall, there is a 75% chance with each pregnancy the child will **not** have JLNS. If a carrier parent demonstrates symptoms of long-QT syndrome, though, any siblings of an affected child should also be offered appropriate cardiac testing.

Management

The diagnosis of JLNS is made clinically. Genetic testing is available for confirmation of diagnosis, and carrier testing and prenatal diagnosis if a mutation is identified in an affected family member.

Hearing interventions appropriate for severe to profound sensorineural hearing loss should be discussed with the family and properly implemented. Hearing loss can be successfully treated with cochlear implantation. Should a child need a bipolar pacemaker as an intervention for cardiac problems, the cochlear implant will **not** interfere with its function.

The main goal in managing JLNS is to prevent episodes of syncope, cardiac arrest, and sudden death. As such, regular appointments with a pediatric cardiologist are vital. Beta-adrenergic blockers are often the first step in treatment, but the efficacy of these drugs is doubtful and they may not prevent symptoms in patients with JLNS. More aggressive measures like a pacemaker or even an implantable cardioverter defibrillator may be necessary. Studies have suggested that individuals with JLNS may be divided into “high risk” and “low risk” patients in regards to cardiac problems. High risk patients have a Q-T interval at or above 550 msec, a history of at least one syncopal episode in the first year of life, a *KCNQ1* mutation, and are males under 20 years of age. These patients would gain the most benefit from early implantation of a cardioverter defibrillator. Low risk patients should have an external defibrillator available.

Any drugs that may cause even greater prolongation of the Q-T interval should be avoided. The same can be said for activities that may precipitate syncope, such as competitive sports (especially swimming), amusement park rides, scary movies, or anything that may trigger shock or intense, sudden emotion. Many family members choose to be trained in CPR in case of syncope or cardiac arrest.

A scheduled appointment with a geneticist and/or genetic counselor is recommended.

Resources for Families

Statewide Genetics Program

Phone: 608-267-7148

Fax: 608-267-3824

Email: meyeram@dhfs.state.wi.us

Wisconsin First Step Hotline

Phone: 1-800-642-7837 voice/TTY

Website: www.mch-hotlines.org

Wisconsin Office for Deaf and Hard of Hearing

Phone: 1-608-266-3118 voice/TTY

Website: www.dhfs.state.wi.us/sensory

Regional Children and Youth with Special Health Care Needs Centers

Centers in Green Bay, Wausau, Milwaukee, Madison, and Chippewa Falls

Website: http://dfhs.wisconsin.gov/DPH_BFCH/cshcn/index.HTM

WI Chapter of Families for Hands & Voices

Phone: (920) 437-7370

Website: www.handsandvoices.org

Parent-to-Parent of Wisconsin

Phone: 1-888-266-0028

Email: mathea@shsmh.org

National Organization for Rare Disorders (NORD)

www.rarediseases.org