

## Clinical Forum

# Congenital Hearing Loss in Jervell and Lange-Nielsen Syndrome

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### Abstract

Jervell and Lange-Nielsen syndrome is an autosomal recessive hereditary condition that presents with cardiac abnormalities characterized by a prolonged Q-T electrocardiographic pattern and congenital severe-to-profound auditory deficits. This paper describes the auditory history of twin boys born out of consanguinity and diagnosed with this syndrome. Both infants were products of the neonatal intensive care unit (NICU) and failed initial ABR screening. Diagnostic evaluation demonstrated profound hearing loss and developmental delays for each infant. Because sudden death is a consequence, audiologists are advised to recognize signs and symptoms associated with this syndrome.

**Key Words:** Jervell and Lange-Nielsen syndrome, consanguinity, congenital hearing loss, prolonged Q-T interval, cardioauditory syndrome, hearing disorders, child

**J**ervell and Lange-Nielsen (1957) syndrome is an autosomal recessive condition, characterized by cardiac arrhythmias and syncopal (fainting) attacks that are thought to be triggered by emotional or physical stress. Loss of consciousness requiring cardiorespiratory resuscitation is common. In its severest state, death, attributable to ventricular fibrillation and asystole, follows.

The electrocardiogram (ECG) is a graphic recording of the electric current produced by heart contraction that display voltage variations resulting from the depolarization and repolarization of the heart muscle over time. The ECG is described in terms of P, Q, R, S, and T waves. Alterations in wave patterns are indicative of abnormalities in the cardiac cycle.

Cardiac defects in this syndrome are characterized by a prolonged Q-T interval and, inverted or biphasic Twave patterns of the electrocardiogram (ECG) (Jung, 1989). The Q-T interval represents the depolarization and repolarization of the heart muscle. The Q-T interval varies with the cardiac rate and should not be greater than 0.43 second for rates above 60 beats per minute. Prolonging this interval makes the heart muscle more vulnerable to ventricular tachycardia and/or ventricular fibrillation and thus, the risk of syncope or sudden death. When congenital sensory hearing loss is associated, the condition has been termed Jervell and Lange-Nielsen syndrome. Consanguinity has been reported in 21 percent of those with congenital deafness (Mosavy and Shafegh, 1976). In the absence of deafness, the disorder is referred to as the Romano-Ward syndrome having an autosomal dominant inheritance (Romano et al, 1963; Ward, 1964). To date, approximately 500 documented cases of prolonged Q-T syndrome have been reported (Corcos et al, 1989). Although not restricted to children, the majority of cases have been reported prior to 10 years of age. This case study describes a family with prolonged Q-T syndrome of which twin off-

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spring have been positively diagnosed with Jervell and Lange-Nielsen syndrome.

### NATURE OF THE SYNDROME

**A**lthough the pathogenesis of the prolonged Q-T interval in patients is unresolved, ventricular premature systoles, ventricular tachycardia and fibrillation, supraventricular arrhythmias, and ventricular asystole have been described (Mosavy and Shafegh, 1976). Bradycardia may also be the presenting sign.

The Jervell and Lange-Nielsen syndrome has been associated with abnormality in vascularization of the sinus node, myocardial metabolism, and to the effects of adrenergic stimulation (Schwartz et al, 1975). However, there appears to be support for a direct relationship between the prolonged Q-T patterns and the sympathetic nervous system. Schwartz and colleagues (1975; 1985) have proposed that the pathogenetic mechanism is an imbalance in sympathetic innervation decreasing right-sided cardiac activity. Others (Ward, 1985; Till et al, 1988), however, support the contention that the sympathetic nervous system may only trigger intrinsic abnormalities at the myocardial cell level. Beta-blocking agents such as propranolol have been used most frequently and effectively. Sympathetic heart antagonists have reduced the mortality of the syndrome significantly. Other antidysrhythmic drugs such as phenytoin have also been used to prevent ventricular dysrhythmias.

### AUDITORY DEFICITS

**J**ervell and Lange-Nielsen syndrome, commonly called the surdcardiac syndrome (*surdus* from Latin, meaning deaf), is characterized by congenital severe-to-profound bilaterally sensory hearing loss, although milder states of hearing sensitivity have been reported (Corcos et al, 1989). The disorder affects about 0.3 percent of the congenitally deaf (Schwartz et al, 1975) with the cases of deafness representing 6 to 30 percent of all the patients with a prolonged Q-T pattern (Schwartz et al, 1975; Moss et al, 1985).

Friedmann et al (1966) reported the auditory pathology findings of two young adults with Jervell and Lange-Nielsen syndrome. They found a widespread degeneration of the cochlea and vestibular mechanisms. The tectorial mem-

brane was detached and adherent to the surface of the spiral limbus. Reissner's membrane was collapsed and bonded to the stria vascularis, tectorial membrane, and to remnants of the organ of Corti.

The audiogram typically displays a severe-to-profound hearing loss at higher audiometric frequencies. Middle ear anomalies have not been consistently reported. The speech and language problems are secondary to deafness and vary with its severity (Jung, 1989).

### CASE REPORTS

**T**his case presentation describes the auditory pattern of two twin brothers with confirmed Jervell and Lange-Nielsen syndrome. The 23-year-old mother has also been positively diagnosed with a prolonged Q-T but has refused auditory assessment. Nonetheless, a review of the mother's hospital chart indicates that family members have said that during conversation she "ignores people," and the maternal grandmother reports that the mother had "learning trouble" in school. The 25-year-old father has no apparent symptomatology. The parents are first cousins.

In addition, the twin brothers have a 6-year-old half-sister who was diagnosed with a prolonged Q-T pattern. The child has been adopted, and no further medical history is available. Finally, there has been the recent family addition of a full-term female with no pre- or perinatal complications (currently 4 months of age). Initial evaluation shows no symptoms of cardiac abnormality, a normal EEG, and normal hearing sensitivity as evaluated with auditory brainstem response (ABR) measures. The twin boys have been placed in foster care. The youngest infant is currently in the care of her parents.

#### Case 1

WD is an 18-month-old male infant diagnosed with Jervell and Lange-Nielsen syndrome. The child was a 36-week gestational age delivery weighing 2180 grams. Apgar scores were 1 and 5 at 1 and 5 minutes respectively. The infant spent 2 weeks in the neonatal intensive care unit (NICU) with a history of birth asphyxia, infantile spasm, sepsis, and respiratory distress syndrome receiving mechanical ventilation. Because of bradycardia, the infant underwent an ECG with positive evidence of prolonged Q-T patterns (0.51 seconds). Flash visual-evoked

potentials were within normal limits save for a mild intereye asymmetry of uncertain significance.

Because of the admission to the NICU, an ABR screening was performed resulting in bilateral failure. At 2 months of age, the infant was again tested with no observed ABR responses at maximum intensities. Under sound field testing, behavioral observation audiometry failed to demonstrate any startle responses at the limits of the audiometer using speech, white noise, and narrow bands of noise. The child has received two post-auricular hearing aids although longitudinal evaluation has not confirmed productive functional use. The child has been diagnosed in the mentally retarded range and according to psychological evaluation is developmentally delayed about 9 months.

#### Case 2

SD, twin brother of Case 1 is an 18-month-old male with Jervell and Lange-Nielsen syndrome. This preterm infant weighed 1960 grams at birth and was admitted to the NICU for an 8 week period with a history of apnea, bradycardia, prolonged Q-T syndrome (Q-T interval of 0.52 seconds), gastroesophageal reflux, and inguinal hernia. Apgar scores were 8 and 9 respectively. An EEG was recorded and showed grossly abnormal high voltage, slow asynchronized background with innumerable superimposed multifocal and generalized spikes suggesting diffuse/multifocal epileptogenic encephalopathy. A left-sided weakness has been documented noted to a flattened head secondary to continued intrauterine pressure from his larger brother.

The initial ABR screen was conducted at 1 week of life resulting in bilateral failure. A diagnostic ABR evaluation followed at 41 weeks post-conceptual age. No reproducible ABR responses were observed at maximum intensities (95 dB nHL). Behavioral testing has reaffirmed the initial diagnosis of profound bilateral sensory hearing loss. Unfortunately, several hospital admissions prevented early intervention for hearing impairment. The infant was fitted with bilateral post-auricular amplification at age 16 months, but as in the twin brother, to date, no improvement in communicative skills has been recorded.

Currently, both twin boys are enrolled in a preschool hearing impaired program, are receiving infant stimulation through occupational and

physical therapy, and are on medication to control cardiac arrhythmia.

#### COMMENT

Because of the prevalence of deafness and possible mortality associated with this syndrome, audiologists and other professionals working with the infant and pediatric deaf population must learn to recognize characteristic signs and symptoms of this syndrome. Further, an ECG screen is recommended for any congenitally deaf infant. Abnormal ECG patterns are potential life-threatening conditions and must be diagnosed early; as well, cardiologists should refer patients with a prolonged Q-T pattern for a formal hearing evaluation. Early diagnosis and management strategies are important facets in the aural rehabilitation of infants diagnosed with Jervell and Lange-Nielsen syndrome. Appropriate genetic counseling should be included in the work-up.

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