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A MALADY OF THE HEART AND EARS: A RARE CASE OF JERVELL AND LANGE NIELSON SYNDROME - CASE REPORT

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ABSTRACT
Jervell and Lange-Nielsen syndrome (JLNS) is a autosomal recessive disease,characterized by profound congenital sensorineural deafness and a prolonged QTc interval (greater than 500 milliseconds). Sudden cardiac deaths have been reported in morethan 25% of JLNS patients. JLNS can occur without any cardiac symptoms. Pharmacological management consists of β-blockers. We are reporting a 5 year old child with JLNS without any cardiac symptoms and his prolonged QT_c interval responded to propranolol.

KEY WORDS:- Jervell and Lange-Nielsen syndrome (JLNS)., Prolonged QTc interval, Propranolol

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Case Report

A 5year old male child was brought with history of delayed speech and decreased hearing first noticed since 1 year of age. He was born out of a third degree consanguineous marriage to parents with normal hearing. His birth history was unremarkable, and had achieved milestones appropriate for age in the motor and social fields. There was positive family history of deafness in mother's cousin. He had been using hearing aids for 1 year but with no benefit. Hearing evaluation in the form of Pure tone audiometry, Evoked Potential, Conditioned Play Audiometry were done which showed bilateral severe to profound hearing loss.CT of temporal bone was normal. His vitals were within normal limits and he had no external physical abnormalities. During pre-anaesthetic evaluation and fitness for Cochlear Implantation [CI], ECG disclosed QT prolongation with $QT_c$of 569msecs[Fig 1A]. His serum calcium, magnesium,sodium and potassium levels were normal.2D Echocardiography was normal. Child had no previous episodes of syncope, convulsions, palpitations, or breathlessness. A diagnosis of Jervell and Lange Nielson syndrome was made. ECG of both the parents were assessed to be within normal limits.Counselling was done andhe was started on oral Propranolol at 1mg/kg/day 8^{th} hourly. Cochlear implantation was deferred. Warning signs were explained and a list of medications to be avoided was provided, and the child was on follow up. After 2 months of treatment, repeat ECG was done which showed further prolongation of $QT_c$to 629msecs and dose of Propranolol was increased to 1.6mg/kg/day 8^{th} hourly. After a month of the new treatment, ECG showed improving $QT_c$of 483msecs[Fig 1 B]. Child remained asymptomatic throughout the course of treatment. At present he is on Tab Propranolol 10mg 1tab-1tab-1/2tab. Child was subsequently operated and a cochlear implant inserted successfully without any complications under nitrous oxide and isoflurane anaesthesia after stabilization of his condition.

Discussion

Jervell and Lange-Nielsen syndrome (JLNS) is characterized by profound congenital sensorineural deafness and a prolonged QTc interval (greater than 500 milliseconds)[1]. It is a autosomal recessive disease and 90% of cases are caused by KCNQ1 gene mutations [1].The prevalence of JLNS is about 1/200,000 to 1/1,000,000 children

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[1,2]. It is one of the most severe forms of long QT syndrome (LQTS) [2]. Anaesthesia can trigger life-threatening arrhythmiasduring CI surgery[1].Yue Qiuet al reported convulsion and life-threatening cardiac arrhythmias in a child at the end of the CI surgery[1]. Sudden cardiac deaths have been reported in morethan 25% of JLNS patients[1]. JLNS can occur without any cardiacsymptoms[1]. Triggers of cardiac arrthythmias in children include exercise, emotion, swimming, auditory stimuli, anaesthesia, and fever[1]. Clinical symptoms like syncopal attacks occurs mostly in early childhood during periods of stress/exercise and carry high risk of sudden cardiac death[2]. In JLNS, 50% of patients experience event by the age of 3 years and 90% of have developed symptoms by 18 years of age[2]. Adadi et al reported multiple episodes of syncope in a 10-year-old Moroccan boy[2]. However our child was asymptomatic. Anaesthesia is very important. Asymptomatic patients may develop Ventricular tachycardia for the first time in the operation theatre[3]. Goyal et al reported JLNS in a child who presented with refractory epilepsy[4]. They suspected cardiac cause as the cause of seizures because the child was hypotensive and pulseless during the episode of seizures. They could stop antiepileptic drugs and child responded to β-blocker therapy[4]. ECG screening of family members also revealed a QT interval more than required for the diagnosis of LQTS but they were asymptomatic[4]. However QT interval inECG of parents of our child was within normal limits. Management of JLNS consists of β-blockers, implantable cadioverters and defibrillators for the cardiac condition[4]. Among the beta blockers, Propranolol is the standard recommendation & other being Metoprolol[4]. In LQTS,β-blockers significantly decrease the risk of sudden death[4]. Our childQT interval increased to 629 mseconds with 2 months of Propranolol at 1mg/kg/day.Therefore dose increased to 1.6mg/kg/day.After onemonth,ECG showed improving $QT_c$of 483msecs.However our child never had any cardiac symptoms.Educating the parents regarding precipitating factors which increase QT interval like competitive sports, amusement park rides, scary movies, jumping into the cold water is very important[4]. We also counselled regarding the risk factors.Along with this cardiopulmonary resuscitation training of family members is also necessary [4].

To conclude, in a child with congenital sensorineural deafness,an ECG must be taken to detect prolonged QT interval so as to take the necessary precautions to prevent precipitation of cardiac arrythmias, and to practice utmost vigilance during CI surger

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LEGEND

FIG 1 A:- ECG in JLNS with prolonged QTc interval.

FIG 1 B :- QTc interval after 3 months of propranolol therapy.

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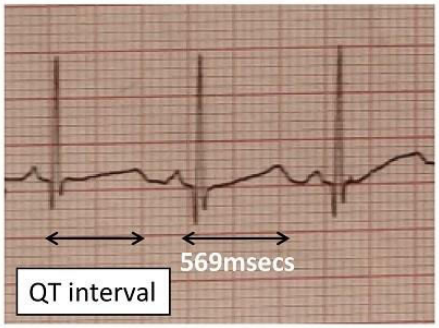
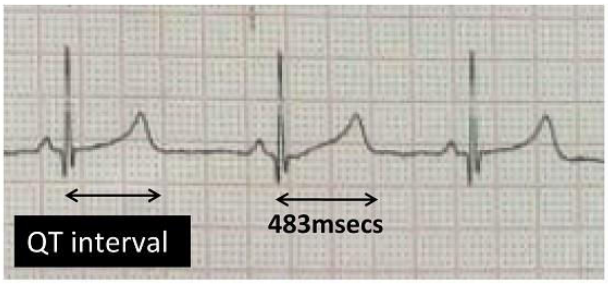
A



At the time of diagnosis

B



After 3 months of Propranolol