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Syndromic deafness-prevalence, distribution and hearing management protocol in Indian scenario

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ABSTRACT

Background: The estimated prevalence of Sensory Neural Hearing Loss (SNHL) in patients less than 18 years of age is 6 per 1000. Roughly 50% of cases of congenital SNHL can be linked to a genetic cause, with approximately 30% being syndromic and the remaining 70% being non-syndromic. The term "syndromic" implies the presence of other distinctive clinical features in addition to hearing loss. The aim of our study was to find the distribution of various Syndromic associations in patients with profound deafness, presented at Madras ENT Research foundation, Chennai and to formulate a management protocol for these patients and to discuss in detail about the clinical features of commonly encountered syndromic deafness.

Materials and methods: Our retrospective study was aimed at describing the various Syndromic associations seen in patients with congenital profound deafness. Information was collected from the medical records. At our centre all patients undergo a comprehensive evaluation. The distribution, etiological factors and management protocol for various syndromes are here presented.

Results: Out of 700 patients with congenital profound deafness all patients with Syndromic associations (n = 35) were studied. 5% of profoundly deaf candidates were found to be syndromic. Most common syndrome in our series was found to be congenital rubella syndrome followed by Jervell and Lange-Nielsen syndrome.

Conclusion: Congenital deafness is an associated feature of many syndromes. Detailed history taking with comprehensive evaluation is mandatory to rule out the associated syndromes. Diagnosis must be confirm by a genetic study. Multidisciplinary approach is essential for appropriate diagnosis and management.

1. Introduction

1.1. Background

The estimated prevalence of Sensori Neural Hearing Loss (SNHL) in patients under 18 years of age is 6 per 1000 [1], making it one of the leading causes of childhood disability and a common reason for otolaryngology referrals. Approximately 50% of cases of congenital SNHL can be linked to a genetic cause, with approximately 30% of these being syndromic and the remaining 70% being non-syndromic [2].

The term "syndromic" implies the presence of other distinctive clinical features in addition to hearing loss, and to date 300 syndromic forms of hearing loss have been described [3]. In many syndromes, hearing loss is an inconstant feature, and a complete review of all syndromes associated with hearing loss is beyond the scope of this report.

Cross-sectional imaging is now routinely performed in these patients because it provides important information about possible etiologies of the hearing loss, defines the anatomy of the temporal bone and the central auditory pathway, and identifies additional intracranial abnormalities.

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1.2. Objectives

The aim of the study is to estimate the distribution of various Syndromic associations in patients with profound deafness reported to our institution, which is a tertiary referral centre that receives patients from the entire country, and to describe a management protocol for candidates with hearing impairment in Syndromic deafness.

2. Materials and methods

This is a retrospective review of 700 patients with congenital profound deafness treated at our institution Madras ENT Research Foundation (MERF) over a period of 14 years (January 1999 to December 2013).

All candidates for cochlear implantation underwent a comprehensive medical history, essential ENT, General and systemic examination, routine blood tests [including thyroid function, renal function and serology for Toxoplasma gondii, Rubella virus, Cytomegalovirus and Herpes simplex virus infection (TORCH)], chest X rays, electrocardiogram (ECG), echocardiogram, abdominal ultrasound and radiological evaluation such as high resolution computed tomography and magnetic resonance imaging of inner ear and brain. Findings of evaluation by pediatrician, ophthalmologist, cardiologist, clinical psychologist and occupational therapist were also collected. From the collected data the associated syndromes were identified. Challenges encountered in these patients during surgery and post-operative habilitation were described.

3. Results

Out of the 700 patients referred to our institution with SNHL, 35 patients (5%) were found to have syndromic association. Among the 35 patients, 28 were born to consanguineous parents (80%) (Table 1).

When syndromic associations were suspected after a detailed medical history was taken, systemic examination was performed, and apart from the routine investigations, additional relevant investigations such as ultrasound abdomen showing single kidney, echocardiogram to know the cardiac defects, MRI brain to see corpus callosal agenesis, Thyroid function test to know the thyroid hormone status were performed in these 35 patients. The patient characteristics suggestive of a syndromic association are reported on Fig. 1. Based on these additional investigations, various syndromic associations were identified and are presented in Fig. 2.

The most common syndrome in our series is Congenital Rubella Syndrome.

Although many of these syndromes do not usually demonstrate gross inner ear anomalies by imaging, there are several in which inner ear malformations are a common and sometimes defining feature (Table 2).

3.1. Management protocol

Out of the 35 patients with SNHL and a syndromic association, 34 patients underwent cochlear implantation uneventfully as per standard protocol, implanted by the same senior surgeon using a standard technique. None of the patients of our series encountered any complications intra operatively or post operatively. The cochlear implantation was not performed in one patient with Branchio-oto-cardio skeletal syndrome because the patient was referred to us at the age of 14 years.

The following was the management of the patients with SNHL and syndromic associations:

3.2. Branchio-oto-renal (BOR) syndrome

Both of our patients underwent cochlear implantation with nucleus contour advanced electrode. Both the patients had single functional kidney hence needed no additional intervention from the renal stand point. Bilateral branchial cyst excision was done.

3.3. Branchio-oto-cardio-skeletal (BOCS) syndrome

This patient presented to us at 14 years of age and since desirable outcomes after cochlear implantation is not possible due to neural plasticity at this age implantation was not done. The patient underwent excision of bilateral branchial fistula later. She also had mitral valve prolapse that was managed conservatively.

3.4. CHARGE syndrome

Both of our patients underwent cochlear implantation with nucleus contour advanced electrode. Ophthalmologic, pediatric and cardiac opinions were sought and concurrent conditions managed appropriately.

3.5. Pendred Syndrome

Cochlear implantation was done with nucleus contour advanced electrode. The patient was under the care of an endocrinologist. Her thyroid function test was found to be normal and so regular follow up of thyroid status alone was advised.

3.6. Goldenhar syndrome

The child had patent ductus arteriosus that was previously ligated and infective endocarditis prophylaxis was given and hemodynamics was maintained during surgery. The patient was implanted with the MedEL pulsar device (Innsbruck Austria) with opus II speech processor.

3.7. Alport syndrome

Three patients with Alports syndrome were implanted with the MedEL pulsar device (Innsbruck Austria) with opus II speech processor. All three patients had undergone renal transplantation prior to cochlear implantation.

3.8. Jervell Lange Nielsen Syndrome

All six patients were implanted with the MedEL pulsar device (Innsbruck Austria) with opus II speech processor. Pre – operatively infective endocarditis prophylaxis was given. Serum potassium levels were measured and maintained within normal limits since hypo – kalemia or hyper-kalemia would precipitate an arrhythmia. Beta blocker therapy was started pre – operatively. Intra – operatively, intra venous magnesium and beta blockers (Metoprolol) were used, whenever necessary for correction of the arrhythmia. External temporary pacing was done pre – operatively to correct any arrhythmia during surgery. Monopolar electrocautery was avoided. Drugs with a propensity to prolong QTc interval were avoided. Defibrillator and antiarrhythmic drugs were kept available during the procedure. Acetylcholinesterase inhibitors (Neo stigmine) were not used for reversal of neuromuscular blockade and the patients were allowed to recover from the anesthesia spontaneously.

Post operatively patients were monitored continuously in cardiac intensive care unit for 48 h and, demand pacing was continued,