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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.

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## 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,

**Table 1**
Demographic and clinical characteristics of different hereditary syndromes with SNHL.

| Patient | Age in years | Sex | Positive clinical features | Syndrome |
|---|---|---|---|---|
| 1 | 1 | M | Single functional kidney, Bilateral branchial cyst,[a,b] | BOR |
| 2 | 5 | M | LBW, Repeated UTI, Single functional kidney, Bilateral branchial cyst,[a] | BOR |
| 3 | 14 | F | Short Stature, Skeletal Deformities, Bilateral branchial fistula, MVP,[a] | BOCS |
| 4 | 1.5 | M | Short Stature, Skeletal Deformities, Coloboma,[a,b] | CHARGE |
| 5 | 4.5 | M | Repeated RTI, Coloboma, Hypoplastic semicircular canals,[a] | CHARGE |
| 6 | 3 | F | Neck Swelling, Euthyroid goiter,[a] | PENDRED |
| 7 | 5 | M | Previous cardiac interventions, Breathlessness, Skeletal Deformities, LBW,[a] | GOLDENHAR |
| 8 | 2 | M | Repeated UTI, Renal Transplantation,[a] | ALPORT |
| 9 | 6 | F | Renal Transplantation | ALPORT |
| 10 | 5 | M | Renal Transplantatio[a] | ALPORT |
| 11 | 1 | M | Sudden drop attacks, Prolonged QTc Interval,[a] | JLN |
| 12 | 2.5 | M | Family H/O sudden death, Prolonged QTc Interval,[a] | JLN |
| 13 | 4 | M | Prolonged QTc Interval | JLN |
| 14 | 3 | F | Sudden drop attacks, Prolonged QTc Interval | JLN |
| 15 | 5 | F | Family H/O sudden death, Prolonged QTc Interval,[a] | JLN |
| 16 | 6 | M | Prolonged QTc Interval,[a] | JLN |
| 17 | 2 | M | Visual disturbances, Retinitis pigmentosa,[a,b] | USHERS |
| 18 | 3.5 | F | Retinitis pigmentosa,[a] | USHERS |
| 19 | 5 | F | Visual disturbances, Retinitis pigmentosa | USHERS |
| 20 | 4.5 | F | Retinitis pigmentosa,[a,b] | USHERS |
| 21 | 2 | M | Retinitis pigmentosa,[a] | USHERS |
| 22 | 1.5 | M | Maternal rubella, Breathlessness, PS,[a] | RUBELLA |
| 23 | 5.5 | M | Maternal rubella, Failure to thrive, Previous cardiac interventions, PDA,[a] | RUBELLA |
| 24 | 4 | M | Maternal rubella, Previous cardiac interventions, PDA,[a] | RUBELLA |
| 25 | 2.5 | F | Maternal rubella, Breathlessness, PS, Family history of HOH | RUBELLA |
| 26 | 6 | F | Maternal rubella, Previous cardiac interventions, PDA,[a] | RUBELLA |
| 27 | 5 | M | Maternal rubella, Repeated RTI, PS,[a,b] | RUBELL[a] |
| 28 | 3 | F | Maternal rubella, LBW, PS, PDA,[a,b] | RUBELLA |
| 29 | 1.5 | F | Maternal rubella, Previous cardiac interventions, PDA,[a] | RUBELLA |
| 30 | 4 | M | Maternal rubella, Breathlessness, VSD, PFO, Family history of HOH | RUBELLA |
| 31 | 5.5 | F | Maternal rubella, Breathlessness, PS,[a,b] | RUBELLA |
| 32 | 3 | M | Maternal rubella, Convulsions, PDA,[a,b] | RUBELLA |
| 33 | 2 | M | Maternal rubella, Previous cardiac interventions, PS, PDA,[a] | RUBELLA |
| 34 | 5.5 | F | Delayed Mile Stones,[a] | CHUDLEY |
| 35 | 6 | M | Delayed Mile Stones | CHUDLEY |

Abbreviations used in Table 1 and Fig. 1: **LBW**, Low Birth Weight, **RTI** Respiratory Tract Infection; **UTI**, Urinary Tract Infection; **MVP**, Mitral Valve Prolapse; **PS**, Pulmonary Stenosis; **PDA**, Patent Ductus Arteriosus; **VSD**, Ventricular Septal Defect; **PFO**, Patent Foramen Ovale; **BOR**, Branchio Oto Renal; **BOCS**, Branchio Oto Cardio Skeletal; **JLN** Jervell and Lange-Nielsen.
[a] Consanguineous marriage.
[b] Family history of hard of hearing.

beta blockers were also administered post operatively. ECG was monitored during switch-on of the Cochlear Implant device. Macrolides and Quinolone antibiotics, anti histamines such as Ebastine, Diphenhydramine were avoided as they may prolong the QT interval.

### 3.9. Congenital Rubella Syndrome

All 12 patients were implanted with either nucleus contour advanced electrode or the MedEL pulsar device (Innsbruck Austria) with opus II speech processor.

One child was operated for infundibular pulmonary stenosis (resection) and right ventricular outflow tract reconstruction was also for the same child. The patient had undergone right endoscopic dacryocystorhinostomy. For patients with pulmonary stenosis intra operatively continuous heart rate monitoring was done and tachycardia was prevented since tachycardia increases the pressure gradient across the stenosis. Use of myocardial depressant such as Halothane was avoided, instead Isoflurane or Sevoflurane was used. Hypoventilation and desaturation during the procedure was avoided. End tidal Carbon dioxide ($ETCO_2$) was monitored. Patients were extubated in fully awake state. Post operatively hypoxia was avoided by administering Oxygen (4 L/min) and analgesics were used to avoid sympathetic drive by pain. Fluid over load was avoided and diuretics were used in case of lung congestion.

### 3.10. Ushers syndrome

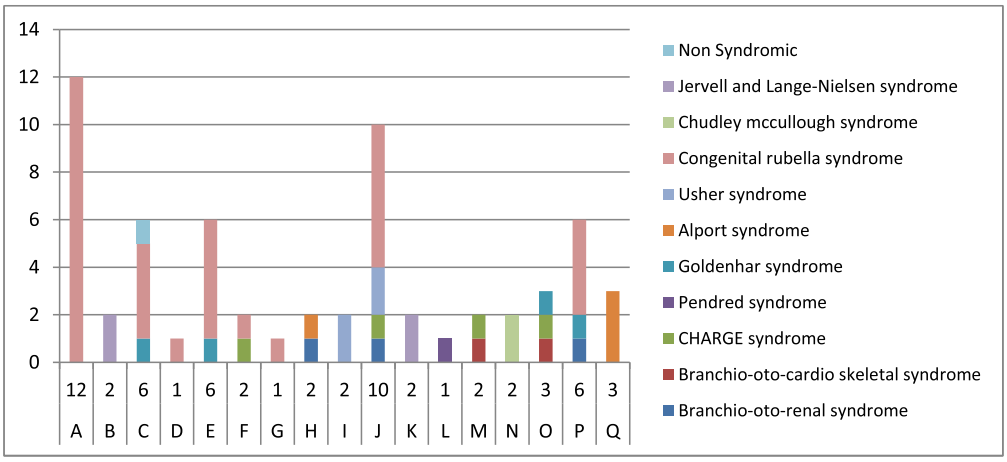
All 5 of them were implanted with either nucleus contour advanced electrode or the MedEL pulsar device (Innsbruck Austria) with opus II speech processor. Early implantation and rehabilitation before visual detoriation was advocated in these candidates.

### 3.11. Chudley-McCullough syndrome

The parents were counseled regarding cochlear implant outcomes and both the patients underwent an uneventful cochlear implantation using a Nucleus contour advanced electrode. Intraoperative electrophysiological assessment was done. Neural Response Telemetry was present with good morphology & amplitude growth and frequency following response was absent.

## 4. Discussion

Our study provides an insight into the various cases of SNHL with syndromic associations commonly referred for cochlear implantation and provides ENT surgeons, a perspective for identifying the syndromic associations and plan the appropriate peri-operative management of these patients. Analyzing the features of various syndromes helped to formulate a protocol for managing complications during anesthesia, explaining expected outcomes to the parents and the need for intense habilitation for certain children.




**Key for alphabets used in the X Axis**

| | Significant Positive history suggesting syndromic associations with deafness | No of affected Children | Syndromes associated |
|---|---|---|---|
| A | Maternal rubella infection | 12 | RUBELLA |
| B | Sudden drop attacks | 2 | JLN |
| C | Breathlessness | 6 | RUBELLA,GOLDENHAR,NON SYNDROMIC |
| D | Failure to thrive | 1 | RUBELLA |
| E | Previous cardiac interventions | 6 | RUBELLA,GOLDENHAR |
| F | Repeated respiratory infection | 2 | RUBELLA, CHARGE |
| G | Convulsions | 1 | RUBELLA |
| H | Repeated Urinary Tract Infection | 2 | ALPORT, BOR |
| I | Visual disturbances | 2 | USHERS |
| J | Family H/O congenital deafness | 10 | RUBELLA,USHER,CHARGE,BOR |
| K | Family H/O sudden death | 2 | JLN |
| L | Neck Swelling | 1 | PENDRED |
| M | Short Stature | 2 | CHARGE, BOCS |
| N | Delayed Mile Stones | 2 | CHUDLEY |
| O | Skeletal Deformities | 3 | BOCS,CHARGE,GOLDENHAR |
| P | Low Birth Weight | 6 | BOR,RUBELLA,GOLDENHAR |
| Q | Renal Transplantation | 3 | ALPORT |

**Fig. 1.** Significant positive history suggesting syndromic associations with deafness.

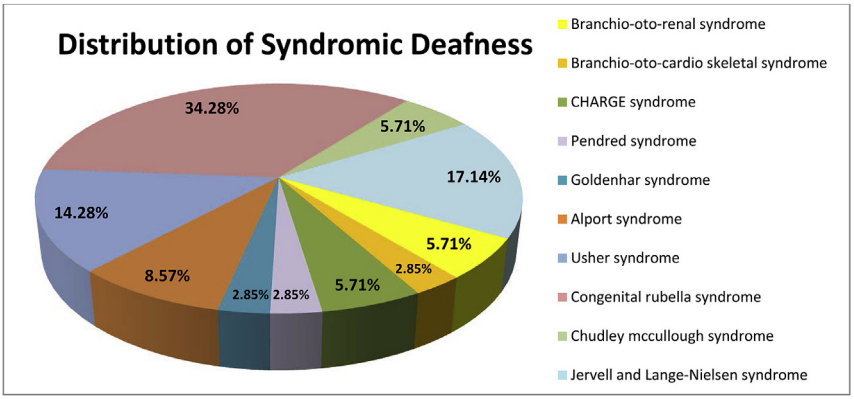



**Fig. 2.** Distribution of selected hereditary syndromes commonly associated with Sensori Neural Hearing Loss (SNHL) in our series.

**Table 2**
Distribution of different hereditary syndromes.

| Syndromes | Number | Percentage |
|---|---|---|
| Branchio-oto-renal syndrome | 2 | 5.71 |
| Branchio-oto-cardio skeletal syndrome | 1 | 2.85 |
| CHARGE syndrome | 2 | 5.71 |
| Pendred syndrome | 1 | 2.85 |
| Goldenhar syndrome | 1 | 2.85 |
| Alport syndrome | 3 | 8.57 |
| Jervell and Lange-Nielsen syndrome | 6 | 17.14 |
| Usher syndrome | 5 | 14.28 |
| Congenital rubella syndrome | 12 | 34.28 |
| Chudley McCullough syndrome | 2 | 5.71 |

### 4.1. Branchio-oto-renal (BOR) syndrome

BOR syndrome is an inherited autosomal dominant disorder and consists of hearing loss, auricular malformations, branchial arch closure defects (preauricular pits and tags), and renal anomalies. Hearing impairment occurs in 70%–93% of individuals with BOR syndrome, with the age of onset varying from early childhood to young adulthood. Hearing loss may be conductive, sensorineural, or mixed and may range from mild to profound [4].

Both of our patients had bilateral profound sensorineural hearing loss with bilateral branchial cyst and single functional kidney.

### 4.2. Branchio-oto-cardio-skeletal (BOCS) syndrome

Features of Branchio-oto-cardio-skeletal syndrome include intrauterine growth retardation, short stature, branchial cyst, sensorineural hearing loss, congenital heart defect, rib and vertebral abnormalities, micromelia, brachymesophalangia, and absence of phalanges [5].

Our patient had bilateral profound sensorineural hearing loss with bilateral branchial fistula and mitral valve prolapse.

### 4.3. CHARGE syndrome

Deafness is observed in approximately 90% of patients, and the hearing loss can be conductive, sensorineural, or mixed [6]. The characteristic inner ear anomaly in CHARGE syndrome is semicircular canal aplasia with associated vestibular dysplasia, which is present in all the patients with the syndrome. Cochlear nerve deficiency with atresia of the cochlear aperture, abnormalities of cochlear partitioning, and anomalies of cranial nerves and olfactory bulbs have also been reported [7].

Our patients had bilateral profound sensorineural hearing loss with Short stature, coloboma, Patent ductus arteriosus, ventricular septal defect and hypoplastic semicircular canals.

### 4.4. Pendred syndrome

Pendred Syndrome is an autosomal recessive disorder characterized by the combination of euthyroid goiter and severe SNHL and represents between 4.3% and 7.5% of all causes of childhood deafness [8].

Our patient had euthyroid goiter and severe SNHL.

### 4.5. Goldenhar syndrome

Goldenhar syndrome is a triad of craniofacial microsomia, spinal anomalies and ocular dermoid cysts. In the ear, it can present with External auditory canal atresia, absent ossicles, facial nerve anomalies and inner ear anomalies with cochlear hearing loss. Goldenhar's syndrome is usually associated with cardiac malformations [9].

Our case of Goldenhar syndrome had right side hemifacial microsomia, facial palsy, pinna deformity, sensorineural hearing loss, cervical vertebral anomaly and ligated PDA.

### 4.6. Alport syndrome

It is a genetic disorder characterized by glomerulonephritis, end-stage kidney disease, and hearing loss [10].

Our patient had renal failure with severe SNHL.

### 4.7. Jervell Lange Nielsen syndrome

This syndrome affects less than one percent of all deaf children [11]. Homozygosity of KVLQT1/KCNE1 gene in chromosome 11p15.5 encodes for a potassium channel 1sK, maintains high potassium in endo-lymphatic fluid in organ of corti and it is expressed in striae vascularis [12].

Jervell Lange Nielsen Syndrome poses an anesthetic challenge. 6 of the 35 patients identified with syndromic deafness were diagnosed to have this syndrome based on ECG criteria (prolonged QTc usually greater than 500 msec – Fig. 3).

### 4.8. Congenital Rubella syndrome

This follows intrauterine infection by the Rubella virus and comprises cardiac, cerebral, ophthalmic and auditory defects [13].

12 of the 35 syndromic patients were diagnosed with congenital rubella syndrome. One of our patients had moderate perimembranous ventricular septal defect, stretched patent foramen ovale and small patent ductus arteriosus was detected. This was treated conservatively. At follow-up, Ventricular Septal Defect (VSD) was found closed, Patent Ductus Arteriosus (PDA) and Patent Foramen Ovale (PFO) was closed. Six patients had pulmonary stenosis as an associated feature.

### 4.9. Ushers syndrome

Usher's Syndrome is an autosomal recessive Syndromic deafness. It is an association of Retinitis pigmentosa with progressive sensorineural deafness. It is the major cause of combined deafness and blindness after childhood [14].

Five patients were diagnosed to have Ushers syndrome. Of these 2 patients had subsequently progressed to near complete loss of vision 4–5 years following their cochlear implantation, but both the patients are in normal auditory verbal communication mode.

### 4.10. Chudley-McCullough syndrome

It is an autosomal recessive condition first reported by Chudley in 1997 [15]. It comprises profound sensorineural hearing loss and hydrocephalus secondary to an obstruction of the foramen of Munro.

In our series two patients had this syndrome. The post-op habilitation was much more intensive than usual and the implantees now have improved response to commands, and improved social interaction with CAP score of 5 and SIR score of 4 in both the patients.

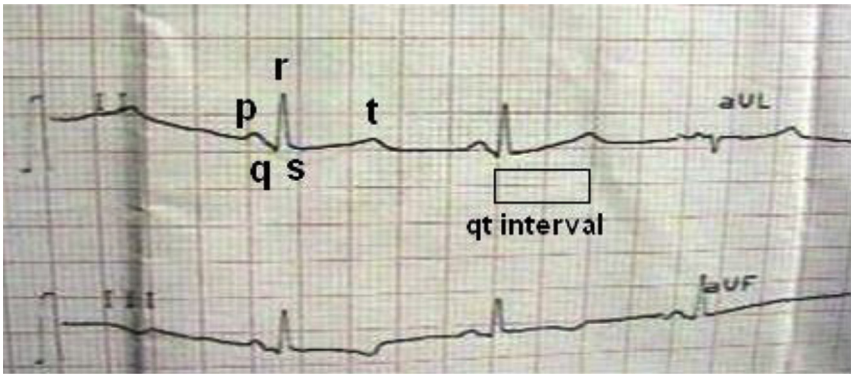



**Fig. 3.** ECG recording showing prolonged QTc interval.

### *4.11. Limitations of our study*

Genetic studies to confirm the diagnosis of syndromic associations were not done. This is a single institutional study but in a country like India with diversity in cultural practices a multicentric study may be needed. The diagnosis and management of other associated conditions were just only mentioned but not discussed in detail.

## 5. Conclusion

Profound deafness is an associated feature of many syndromes. Detailed history taking with comprehensive evaluation is mandatory to rule out the associated syndromes. Most common syndrome in our series was found to be congenital rubella syndrome followed by Jervell and Lange-Nielsen syndrome. Syndromic deafness puts additional burden on the habilitation process after cochlear implantation. A multidisciplinary approach towards the management protocol is essential for Syndromic patients.

Devising a protocol for management of such cases is vital for obtaining the best outcomes in cochlear implantees with syndromic associations. Children with syndromic associations have no contraindication for cochlear implantation. Cochlear implantation is safe in children with syndromic associations provided they are diagnosed early and treated appropriately.

Our article provides an insight in to the various Syndromic issues commonly related to Cochlear implantation and provides cochlear implant surgeons, a perspective for categorizing the problems and plan the appropriate management. Forming such a protocol is a helpful aid for Cochlear implant surgeons worldwide providing them vital guidelines to plan for future implantees.

## Ethical approval

Madras ENT Research Foundation and MERF Institute of Speech and Hearing Ethical Committee approved. Ref. no – 15/2013.

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## Author contribution

Dr.SenthilVadivu – Data Collection, Preparation of Manuscript, Data Analysis.

Dr.Vijaya Krishnan – Data Collection, Preparation of Manuscript, Art work.

Dr.SathiyaMurali – Preparation of Manuscript.

Dr.Kiran Natarajan – Literature review.

Dr.Sudhamaheswari – Literature review.

Prof. Mohan Kameswaran – Study Concept, Manuscript review and interpretation of result.

## Conflicts of interest

No conflicts of interest.

## Guarantor

Dr.Arumugam Senthil Vadivu
Dr.Vijaya Krishnan
Prof. Mohan Kameswaran

## Consent

Fully informed written consent obtained.

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**Appendix-1**

Serial number:

| | | |
|---|---|---|
| 1. | Name | : Age: Sex: |
| 2. | Address | : |
| 3. | H/O Consanguineous marriage | : Yes / No (if Yes _____ degree) |
| 4. | Antenatal history | : Infection / Medication (if Yes, details_____ |
| 5. | Birth history | : Full term / Pre term |
| 6. | Mode of delivery | : Via naturalis / Caesarean section |
| 7. | APGAR Score/ Birth weight | : |
| 8. | Post natal NICU admission | : Yes / No (if Yes, details_____) |
| 9. | H/O cyanotic spells | : Yes / No |
| 10. | H/O sudden drop attacks | : Yes / No |
| 11. | H/O breathlessness | : Yes / No |
| 12. | H/O failure to thrive | : Yes / No |
| 13. | H/O previous cardiac interventions | : Yes / No |
| 14. | H/O repeated respiratory infection | : Yes / No |
| 15. | H/O Convulsions | : Yes / No |
| 16. | H/O suggestive of renal diseases | : Yes / No (if Yes, details_____) |
| 17. | H/O suggestive of thyroid diseases | : Yes / No (if Yes, details_____) |
| 18. | H/O visual disturbances | : Yes / No (if Yes, details_____) |
| 19. | H/O delayed mile stones | : Yes / No (if Yes, details_____) |
| 20. | H/O exanthematous fever | : Yes / No (if Yes, details_____) |
| 24. | Other past medical history if any | : _____ |
| 25. | Other past surgical history if any | : _____ |
| 26. | Family H/O medical illness | : Yes / No (if Yes, details_____) |
| 27. | Family H/O congenital deafness | : Yes / No |
| 28. | Family H/O sudden death | : Yes / No |
| 29. | Immunized for age | : Yes / No |
| 30. | Treatment history if any | : _____ |

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