

Clinical Forum

Hearing Loss in Stickler's Syndrome: A Family Case Study

John Jacobson*
Claire Jacobson†
William Gibson‡

Abstract

Stickler's syndrome is an autosomal dominant hereditary condition that presents with pathognomonic facial features, dysplasia, myopia and auditory deficits. This paper describes the auditory history of a family with Stickler's syndrome. Given the expressive variability, a complete audiologic and otologic evaluation is encouraged when Stickler's syndrome is suspected.

Key Words: Stickler's syndrome, hearing disorders, hereditary

Stickler's syndrome is an autosomal dominant hereditary condition presenting with clinical traits that include cleft palate and hearing loss. Given the small number of patients identified with Stickler's syndrome, a comprehensive auditory study has yet to be completed. Although the literature suggests hearing loss as one clinical entity, its nature and severity are usually not well documented since hearing impairment has not been a primary objective of most investigations.

Stickler's syndrome yields high expressive variability even within the same family (Jung, 1989). This case study describes one such occurrence. The father presents with a true sensorineural hearing loss, one child with a tentative mixed hearing loss (true sensorineural and transient middle ear pathology presumably related to her cleft palate history) and one infant with normal hearing as demonstrated by ABR.

*Departments of Otolaryngology and Audiology, Geisinger Medical Center, Danville, Pennsylvania

†Department of Audiology, Geisinger Medical Center, Danville, Pennsylvania

‡Department of Otolaryngology, Geisinger Medical Center, Danville, Pennsylvania

Reprint requests: John T. Jacobson, Department of Audiology 42-43, Geisinger Medical Center, Danville, PA 17822

NATURE OF THE SYNDROME

Stickler's syndrome is a connective tissue dysplasia of progressive joint and eye abnormalities (arthro-ophthalmopathy, Stickler et al, 1965; Stickler and Pugh, 1967). It is considered an autosomal dominant genetic disorder, producing expressive variability and high but incomplete penetrance (Weingeist et al, 1982; Suslak and Desposito, 1988). Typical symptoms include mild-to-severe axial myopia with total retinal detachment usually in the first decade of life, characteristic orofacial, skeletal, submucous and cleft palate, bifid uvula, and auditory abnormalities. Analogous clinical traits are reported in Pierre Robin sequence, and it has been suggested that approximately half of those with the Pierre Robin anomaly may have Stickler's syndrome (Herrmann et al, 1975). Thus, with the exception of its pattern of inheritance, many clinical studies have not clearly differentiated between the two disorders. To date, the brief history and small numbers of confirmed Stickler's syndrome patients have resulted in scant clinical evidence of its effects on hearing within this disordered population.

AUDITORY DEFICITS

Hearing loss may be a consequence of either a primary sensorineural or childhood conductive deficit (Weingeist et al, 1982; Stickler

and Pugh, 1967; Popkin and Polomeno, 1974). Congenital abnormalities include characteristically low-set pinnae with associated auricular malformations reported in about 10 percent of cases (Smith and Stowe, 1961; Jung, 1989). Primary middle ear anomalies, usually of the malleus and incus, have been attributed to first branchial arch development, whereas sensory pathology may be due to structural malformation rather than sensory receptor development (Igarashi et al, 1976).

The degree and nature of hearing loss varies considerably. Typically, bilateral conductive hearing loss has been reported in about half of all patients secondary to existing palatal abnormalities (Williams et al, 1981; Gould, 1989). The prevalence of congenital mixed or sensory impairment is less frequent, approximating 10 percent (Popkin and Polomeno, 1974; Jung, 1989; Weingeist et al, 1982). However, Liberfarb and Hirose (1982) have reported a much higher prevalence (87 percent) of sensorineural hearing loss in 39 diagnosed patients. In addition, prevalence of loss was associated with increasing age; that is, those (18) under 30 years of age were asymptomatic, only some patients (13) between 30 and 50 years were aware of hearing loss, whereas all (8) of those over 50 years were aware of some hearing loss.

CASE REPORTS

This case study describes the auditory pattern of a family (father and two children) with confirmed Stickler's syndrome, which has been followed at Geisinger Medical Center (GMC). The father was initially diagnosed with a hearing loss at age 2 and both infants were at risk for hearing loss due to family history. Clinical observation and chart notes suggest a history of Stickler's syndrome in four generations of this family. The father has been aided since the initial diagnosis of his hearing loss at age 2.

Case 1

DD is a 31-year-old white male with Stickler's syndrome who presented with high myopia, congenital cataracts, myopic astigmatism of the right eye, amblyopia of the left, and degener-

ative joint disease primarily of the knee. Facial abnormalities included a typical severe hypoplastic nasal septum but no evidence of cleft palate or lip.

Hearing history indicated an early diagnosis of relatively stable moderate bilateral sensorineural hearing loss. Figure 1 presents the audiometric findings at age 4 (11/62) and the most recent audiometric evaluation (4/88). At the time of evaluation, immittance audiometry indicated normal middle ear function.

Although Stickler's syndrome was not described in the literature until after his birth, hospital chart records indicated "the father has poor hearing" and "the father and grandfather have the same shaped nose." Additionally, otologic examination by one author (WG) shortly after birth reported "consider possible familial syndrome of eye and nasal defect. Eyes are quite prominent, exophthalmic and widely spaced. Bridge of nose is practically absent." Thus, physical examination indicated clinical traits suggestive of Stickler's syndrome in two preceding generations.

Case 2

JD is a 28-month-old female identified with congenital Stickler's syndrome and presenting with cleft palate, mid-facial hypoplasia and micrognathia, and mild developmental delays. The child was a 38-week gestational age deliv-

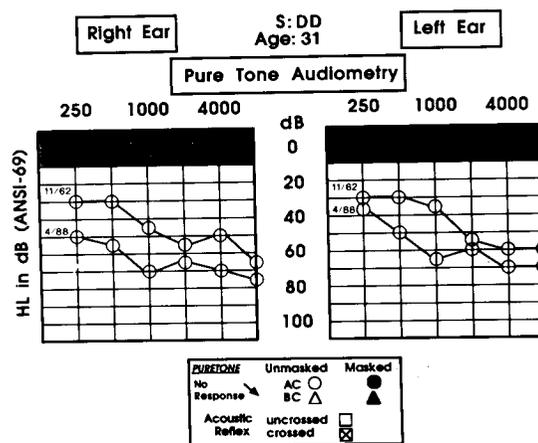


Figure 1 Audiometric findings at age 4 and age 30.

ery, weighing 1980 g. APGAR scores were 3 and 7 at 1 and 5 minutes, respectively. The infant, born in the well baby nursery, was transferred to the NICU for continued care due to pulmonary hypertension and right ventricular hypertrophy. Because of a reactive airway disease and micrognathia, a tracheostomy was performed. The cleft palate has been repaired.

The newborn hearing screening program identified this infant as at-risk for hearing loss due to family history and admission to NICU. Auditory evaluation suggested that this infant presented with a moderate bilateral sensorineural hearing loss. Most recent ABR tracings at age 14 months are shown in Figure 2. Repeatable responses were measured at levels of 45 dB nHL in the right ear and 40 dB nHL in the left. At the time of ABR testing, immittance audiometry showed normal bilateral middle ear function. The presence of normal wave I latencies and Type A tympanograms supported the diagnosis of cochlear pathology. It should be noted, however, that this child has a history of recurrent otitis media with effusion, and ventilation tubes have been placed since the most recent auditory evaluation. This child has been fitted with amplification and is in a pre-school deaf program at GMC. Recent speech and language evaluations show mild delay in both expressive and receptive language.

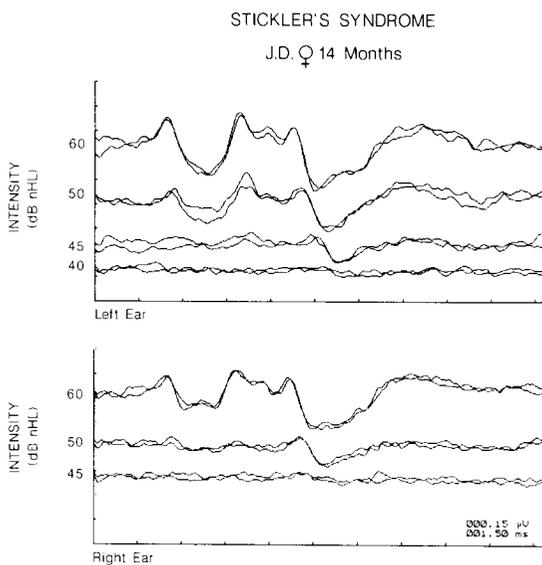


Figure 2 ABR tracings at age 14 months.

Case 3

BD is a 3-month-old male who was identified as at high risk for hearing loss (familial pattern) from the newborn screening program. This full term newborn with an unremarkable birth had APGAR scores of 9 and 9, weighing 3150 g. The physical examination at birth noted typical facial characteristics pathognomonic of Stickler's syndrome although there was no evidence of cleft lip or palate. Figure 3 displays the facial characteristics in Stickler's syndrome as seen in the father (DD) and his children.

Initial ABR screening was conducted within the first week of life. As part of routine protocol in the well baby nursery, he was tested with an automated ABR screener (ALGO-1) and passed bilaterally at 35 dB nHL. However, due to family history and the unknown consequences of emerging hearing loss in Stickler's syndrome, a conventional diagnostic ABR evaluation was conducted at 42 weeks post conceptual age. Test results are illustrated in Figure 4. Bilateral ABR responses were present at 20 dB nHL, suggesting normal peripheral auditory function. Repeat follow-up evaluations are scheduled routinely to confirm normal hearing sensitivity.

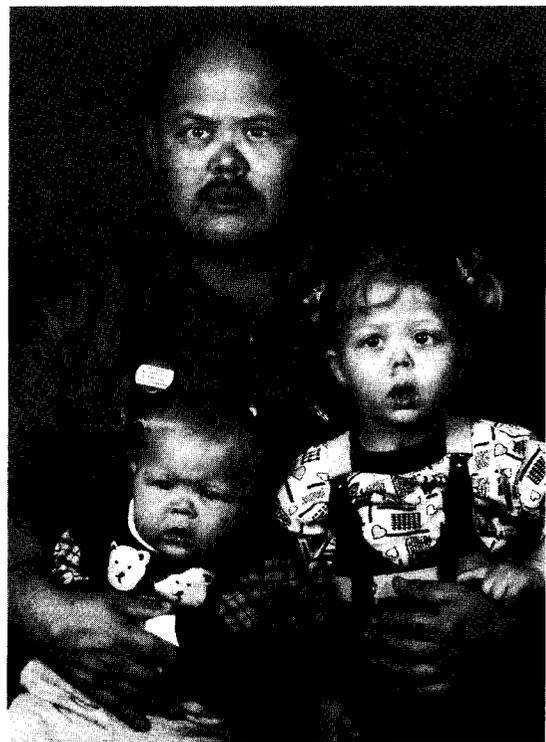


Figure 3 Facial characteristics in Stickler's syndrome as seen in the father and his children.

STICKLER'S SYNDROME

B.J. ♂ 42 wks PCA

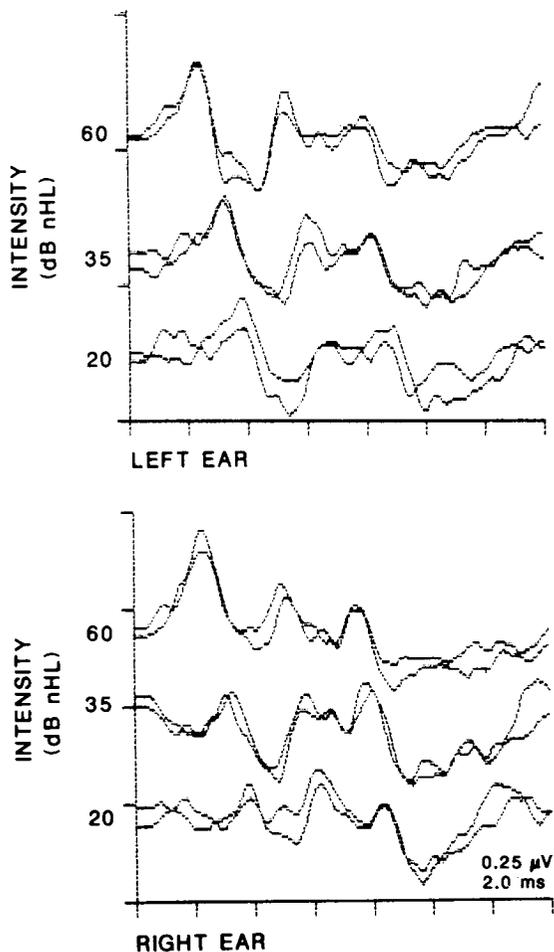


Figure 4 Test results of ABR at 42 weeks post conceptual age.

COMMENT

This family may represent the actual capricious nature of hearing loss in this disorder. Therefore, it is recommended that any adult,

child, or newborn identified with Stickler's syndrome receive a complete audiologic and otologic work-up to rule out possible auditory deficits associated with this condition.

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