Congenital Conductive Hearing Impairment in Noonan Syndrome

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Introduction

In 1963, Noonan and Ehmke reported a number of children with cardiac, pulmonary, and cranio-facial abnormalities. While the phenotype that we now know as Noonan Syndrome had been observed earlier, it was from this time that it was distinguished from other disorders, particularly Turner Syndrome. The Noonan Syndrome patient (Figure 1) presents with short stature, facial anomalies, congenital cardiac defects, skeletal abnormalities, genital malformations, and mild mental retardation. In fact, more than fifty different major and minor anomalies have been observed to occur in Noonan Syndrome (Gorlin, Pindborg, & Cohen, 1976). It is thought to be second only to Down Syndrome in frequency of occurrence among syndromes presenting with multiple congenital anomalies, and it has sometimes been confused with other syndromes (Berman, Desjardins, & Fraser, 1975).

The incidence of Noonan Syndrome has been variously reported to occur from one in one thousand births (Summitt, 1969) to as few as one in twenty-five hundred births (Nora et al., 1974). Virtually all writers on the subject (e.g., Smith, 1976) have included among the stigmata of Noonan Syndrome an observation of low set and/or abnormal auricles (Figures 2 and 3). Curiously, though, with two exceptions (Hopkins-Acos & Bunker, 1979; Wilson & Dyson, 1982), there are no reports in the communicative disorders literature on this syndrome in spite of the fact there are obvious alterations of both the hearing and speech mechanisms.

Smith included "nerve deafness" as an occasional abnormality. But in their encyclopedic work, Konigsmark and Gorlin (1976) claimed that patients with Noonan Syndrome are not deaf. However, they also observed that the syndrome is similar to both the Leopard Syndrome and Turner Syndrome, both of which do frequently incorporate hearing impairment. Nora et al. (1974) did report hearing impairment in 12% of their patients with Noonan Syndrome.

In 1976, Gorlin, Pindborg, and Cohen reported the incidence of several of the stigmata of Noonan Syndrome concluding (as did Smith, 1976) that autosomal dominant inheritance is the likely mode. They reported ptosis of the eyelids in about 60% of the patients, strabismus and saddle nose in at lease 50% of the patients mental retardation in at least 50% of the cases and reported that "sensory-neural deafness and hydrocephaly had been noted rarely." Furthermore, they found tht 40-60% of the cases present with cardiovascular anomalies more frequently on the right side. They also discovered some 15% of patients exhibiting edema of the lower extremities. More than 60% of male patients exhibit cryptorchidism. The short stature of the Noonan patient has long been observed in both males and females along with various alterations of the integumentary system.

Given that the Noonan Syndrome patient presents with a frank anomaly of the facies, it is strange that no one has reported the incidence of conductive hearing impairment. Although, indeed, sensory-neural hearing losses do commonly occur with cranio-facial anomalies (e.g., Waardenburg Syndrome), it seems obvious a priori that any child with an unusual facial appearance should be suspected of having a conductive auditory impairment whether or not there is a probability of sensory-neural loss. Furthermore, since mild mental retardation is said to be a frequent concomitant of Noonan Syndrome, it is equally strange that there is only one published report of speech and language development (Hopkins-Acos and Bunker, 1979) which has come to our attention.

Case Study

The patient reported here (who is the same child reported by Hopkins-Acos and Bunker, 1979) has such frank alterations of the external ear that middle ear anomalies must be strongly suspected (Figures 1-3). He was referred to our Speech and Hearing Center by a pediatric neurologist who cited significant delays of developmental milestones along with the physical characteristics usually associated with Noonan Syndrome. In this case, the report noted bilateral undescended testes, widely spaced nipples, and a cardiac murmur. Routine chromosomal analyses appeared normal. At the age of two years and two months, the patient underwent successful cardiac surgery to correct an atrioseptal defect and pulmonary stenosis.

At 3 1/2 years of age, he exhibited the ability to follow simple commands (such as "shut the door"), but he relied entirely on isolated vowel sounds and gestures for his expressive communication. It appeared that, using these sounds and gestures, he could label objects, request objects and actions, call, protest, and greet people. However, no evidence of repetition with a verbal

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Figure 1. The Noonan Syndrome patient presents with short stature, facial anomalies, congenital cardiac defects, skeletal abnormalities, genital malformation, and mild mental retardation. *Figure 2.* Included among the stigmata of Noonan Syndrome are low-set auricles. *Figure 3.* Included among the stigmata of Noonan Syndrome are abnormally shaped auricles.

model was recorded. He did command objects and attempt to regulate the actions of others but did not engage in any reciprocal interaction with adults or other children. An occupational therapy evaluation concluded that he was functioning one to one-and-one half years below the norm in both his fine and gross motor skills with a larger delay in the gross motor area. Specifically, the occupational therapist noted difficulty crossing the midline and a reduction of muscle tone in both legs.

Following 42 hours of speech/language treatment over a 10 week period, his expressive language was found to consist of gestures which were now paired with spontaneous two or three word utterances. He learned to request objects, protest, call, greet people, and repeat utterances. There appeared to be a corresponding increase in his comprehension abilities as well: he apparently decodes four and five word utterances. It was also reported from the home that transfer in generalization had occured outside the clinic environment.

Otologic examination was negative. The audiologic evaluation is clear; audiograms and tympanograms have been reliably made. It may be seen that his pure tone audiogram (Figure 4) is indicative of conductive impairment while his tympanogram (Figure 5) is shifted downward and very slightly toward the negative side. The audiogram shows an air-bone gap as great as 25 dB.

We are aware of another patient exhibiting Noonan Syndrome who has been evaluated elsewhere. This seven year old, mentally retarded, female patient exhi-

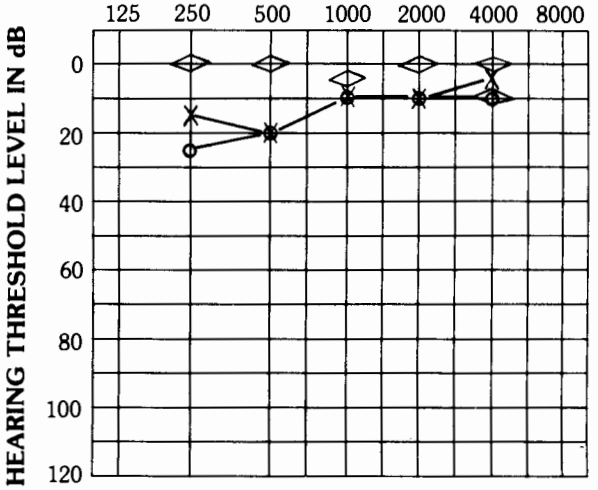

Figure 4. The audiogram of this Noonan Syndrome patient showing an air-bone gap as great as 25 dB.

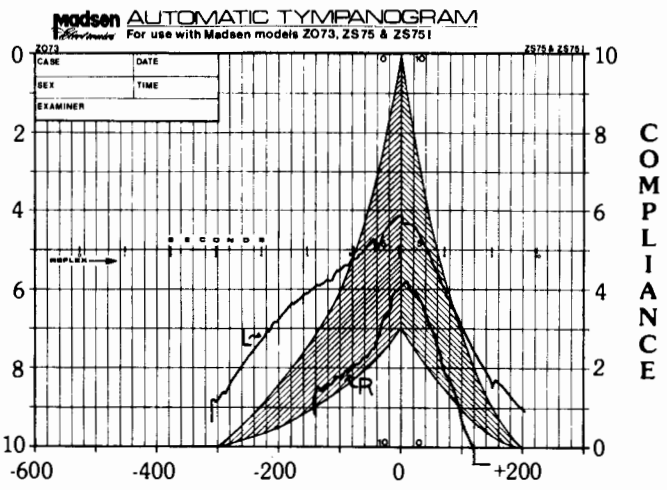

Figure 5. The tympanogram of this Noonan Syndrome patient is shifted downward and slightly toward the negative side.

bited a 30 dB bilateral flat audiogram from 50 HZ - 4000 Hz. She had type B tympanograms bilaterally with static compliance measures below normal. She also had no reflexes at 500, 1000, and 2000 Hz with pure tones, broadband noise, and high-pass or low-pass noises. This patient, too, had pinnae that were unusually formed with large lobes, flattened rim, and folds separating conchae. It is important to observe the pinna alterations in these patients, especially in light of Jaffe's (1978) findings on middle ear anomalies signalled by alterations of the pinnae.

Conclusion

Unfortunately, the data on our patient are equivocal. His poor speech and language development are probably ascribable to his poor intellectual development and not to an expressly auditory deficit. On the other hand, the anatomical alteration as revealed by slightly modified tympanometric configurations should cause the clinician some concern. If we have a child who is clearly physically and mentally deficient, then no amount of auditory deficiency is acceptable. While it is essential that every child be provided maximal use of his auditory system, the necessity for such employment is increased greatly in the child with other handicaps. Furthermore, given a child with an unusual facial appearance and poor mental development, it is too easy to address ourselves totally to these problems and to ignore the probability of auditory difficulties as well.

Clearly, Noonan Syndrome is subject to prevention at primary, secondary, and tertiary levels. The report by Hopkins-Acos and Bunker (1979) is an example of tertiary prevention in the form of habilitation of a particular patient with Noonan Syndrome. Certainly, the fact that this child had had surgery on both the cardiac system and genito-urinary system may be considered a form of secondary prevention. More importantly, however, is the conclusion of Konigsmark and Gorlin (1976) that Noonan Syndrome is due to "possible autosomal dominant inheritance with incomplete penetrance." In other words, the families of Noonan Syndrome patients are good candidates for genetic counseling. Given that they receive the counseling and given that they then elect to have no more children, the incidence and prevalence of Noonan Syndrome should diminish accordingly.

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