A STUDY OF BEHAVIORAL CHARACTERISTICS OF CHILDREN WITH WAARDENBURG'S SYNDROME

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The Waardenburg syndrome, also known as the van der Hoeve-Halbertsma-Waardenburg, Klein syndrome, was first described by Mende in 1926 but clarified by Waardenburg, a Dutch ophthalmologist and geneticist, in 1951 (Cant and Martin, 1967: Konigsmark, 1971). Waardenburg studies the syndrome through fourteen pedigrees and estimated that it accounted for about fourteen percent of the deaf mutes in the Netherlands. The disease showed dominant autosomal inheritance with variable penetrance; it can also occur sporadically as a result of mutations (Marcus, 1968). Although no sex discrimination is shown, congenital deafness occurs more commonly in the male (Chew Kheng Lian *et al.*, 1968). It has been suggested that this syndrome is responsible for 2.3% of the congenitally deaf and that there are some four hundred children in U. S. schools for the deaf who are so affected (Konigsmark, 1969). "Major characteristics of this syndrome include: 1) dominant transmission with variable penetrance, 2) lateral displacement of medial canthi and lacramal points in all affected, 3) broad high nasal root in about 75 percent of those affected, 4) vestibular hypofunction in about 75 percent of those affected, 5) congenital mild to severe unilateral or bilateral neural hearing loss in about one-half of those affected, 6) hyperplasia of medial eyebrows in about one-half of the affected, 7) heterochromia iridium and loss of pigment epithelium of optic fundus in about 25 percent of those affected, 8) white forelock in about 20 percent of those affected, 9) skin pigmentary changes including vitiligo and spotty hyperpigmentation in less than 10 percent of those affected, and 10) hare lip and cleft palate in less than 10 percent of those affected" (Konigsmark, 1971). Premature graying of the hair, eyebrows, and eyelashes may also be characteristic and thought to be connected with or to take the place of the white forelock. The syndrome has also been described as a form of partial albinism (Gellis and Feingold, 1967).

Several studies have emphasized the importance of recognizing the syndrome due to the factor of deafness. Fregonese *et al.*, (1969) discussed two cases of the syndrome placing emphasis on the importance of the disease for the pediatrician. Some research has focused upon ear pathology. Fisch (1959) discussed two types of hearing loss patterns associated with the Waardenburg syndrome. In Type I, there was almost total deafness with some residual hearing at the lower frequencies. In Type II, there was a moderate degree of hearing with a uniform loss in the lower and middle frequencies with an improvement in the higher

tones (Konigsmark, 1971). The inner ear pathology of a profoundly deaf three year old girl who died of pneumonia revealed the organ of Corti to be absent in all coils, and the basilar membrane was slightly thickened and smooth (Fisch, 1959). In the study by Marcus (1968), eighteen patients in a family were tested with only one having a completely normal vestibular function. He also noted that the Waardenburg syndrome has occurred in connection with other syndromes and with albinism. Possibilities of other facial features, such as dystopia, large jaw, upturned nose, and "cupid's bow mouth," have been indicated. He suggests the possibility of various kinds of aplasias occurring in the Waardenburg syndrome involving the osseous and membraneous labyrinths. Jensen (1967) discussed the case of a fifteen year old boy which was regarded as an aplasia of the canal.

The peculiarities associated with the eyes have also been of particular interest. Goldberg (1966) mentioned that the characteristic variations in iris pigmentation are paralleled by the patterns of pigmentation of the fundus. He studied fourteen subjects with the syndrome from seven families and found that fundus pigmentation was abnormal in several patients. Another study compared a case of the syndrome to a case of true hypertelorism (Pryor, 1971). The eyelid deformity in the Waardenburg syndrome gives the appearance of hypertelorism. However, in hypertelorism, all the interocular measurments are increased whereas in the dystopia canthorum found in the Waardenburg syndrome, the distance between the inner canthi of the eyelids is increased while the interpupillary distance is normal (Giacoia and Klein, 1969). Ulivelli and Silenzi (1969) described seven members of one family with the Waardenburg syndrome, four of which also showed pure hypertelorism, which they pointed out as being rare. Stevens (1970) discussed a case in which anterior lenticonus was associated with the syndrome. The interesting feature of the case was the spontaneous rupture of the lens capsule; the lens defects were associated with the Waardenburg syndrome.

Many individual cases and pedigrees have been examined. In a study by Cant and Martin (1967) a family showing all the characteristics of the syndrome in three generations was studied. They reported that the separate features were usually fully expressed but might be present as isolated phenomena. This family was less concerned with the deafness than their appearance and had cosmetic operations as soon as possible. The authors suggested an operation for the correction of dystopia canthi. This physical and cosmetic aspect can be an influencing factor in behavioral characteristics and will be discussed in more detail later. Some researchers have observed a case of Waardenburg's syndrome from birth for several months (Fanaroff and Levin, 1968; Feingold *et al.*, 1967). Rappoport (1970) discussed a "coloured" family showing the syndrome. Bwibo and Mkono (1970) described an African case with the disease and mentioned some other unusual features; they suggested that this might indicate the co-existance of another syndrome. Another case was reported of the syndrome in both husband

and wife; the author mentioned that their chromosomes, dermatoglyphics and intelligence were normal (David, 1971). The study by Roux *et al.* (1970) included twenty-three persons in four generations and discussed, in particular, two individuals with a tumor between both eyebrows and one with a harelip.

As can be seen from the literature, a great deal of research has focused upon the physical traits of the Waardenburg syndrome with very little attention on the behavioral characteristics. The purpose of this paper is to discuss the behavioral aspects of the syndrome.

The research involved a retrospective clinical study of behavioral characteristics of Waardenburg children using the existing school records of the Maryland School for the Deaf and some teachers' judgements. Twenty cases were found from a school population of approximately 2226; this indicates a prevalence of almost one percent of deaf children being affected by the Waardenburg syndrome.

ETIOLOGICAL FACTORS

Since only twenty to fifty percent of those affected by the syndrome have a hearing loss, the possibility exists that a subject with the syndrome could have been deafened by another factor. Only two cases with other possible etiological factors were found. One subject had meningitis at the age of three months; in the other case, there was a report of maternal rubella. In the latter case, the subject was described as having aphasia, but no other differences from the other Waardenburg children were seen. Since frequently there is a lack of desire on the part of parents to admit that the cause of their child's deafness is hereditary, it may be questionable that the rubella and meningitis are significant etiological factors in those cases.

PHYSICAL ANOMALIES, MULTIPLE HANDICAPS

Five subjects (25%) were found to have vision problems; two of these were indicative of ambliopia. Two subjects were thought to have vision difficulties due to the eyelid deformity where the eye may actually be covered by the eyelid. There were three cases with cleft palate, and several subjects had pigmentation disorders normally found in the Waardenburg syndrome, such as a white forelock and different colored iris. One subject had extremely heavy facial markings with the possibility of a nerve paralysis. There was one case of aphasia which has already been mentioned. It has been noted and will be discussed in more detail that some of the physically unattractive aspects of Waardenburg's syndrome affect behavior.

AUDIOMETRIC FINDINGS

Three subjects had a total hearing loss (no response), thirteen were classified as profoundly deaf (90 db loss or more), three were severely hard of hearing (70 to 90 db loss), and one was moderately hard of hearing (50 db loss).

PSYCHOLOGICAL ADJUSTMENT

The Waardenburg subjects, with two exceptions, appeared well adjusted. In the two exceptions, it is felt that environmental, not organic, factors accounted for the differences. The first subject had extremely heavy facial markings, and although a hard worker with at least average ability, he showed uncontrolled hyperactivity which was greatly influenced by other students' reactions to his appearance. He was constantly teased and ridiculed, and peer relations were poor. Since he was already sensitive about his appearance, these additional reactions were ctastrophic.

The second subject came from an extremely poor background; he had an illiterate mother and was hospitalized for a lengthy period before any real attempt at schooling was begun. As a result, his development was extremely bizarre which progressed into emotional problems. He was considered very destructive, irritable and changeable.

These two cases have been mentioned to illustrate the massive effects that environmental reactions to the physical aspects of the syndrome may have, compounding otherwise difficult home environments. A particular cosmetic problem associated with the Waardenburg syndrome is cleft palate. Correction of appearance can be considered, especially in extreme instances of disfiguration. However, it is necessary to recognize that, even with correction, the cosmetic aspect may affect the child's adjustment.

In eleven other cases (55%), hyperactivity, temperamental dispositions, and stubbornness were noted. With the exception of the two previously mentioned subjects, these characteristics would be considered within normal limits and could be related to various factors, such as physical appearance, lack of parental discipline, and being academically ahead of the class.

Six to ten of the twenty subjects have been considered leaders in their classes and/or among the deaf community. These same persons had good communication and language skills and above average IQs. Six of these subjects were given the Vineland Social Maturity Scale and the average of their social quotients was 91. Generally, maturity and social aspects of the subjects were considered normal.

Psychological adjustment of the Waardenburg subjects would be considered good, with the cosmetic and home environment problems being noted. Thirty to fifty percent are leaders within their peer group.

INTELLIGENCE

Waardenburg subjects showed above average intelligence. This is generally true of hereditary deafness, thus suggesting the possibility of a genetic influence in IQ (Vernon, 1969). A review of school records indicated there were IQ test results available for eleven of these children and that the average IQ was 113. Teacher judgments indicated the other children would be judged to be in the normal range (90-109). It can therefore be suggested that persons affected with the Waardenburg syndrome would most likely have above average intelligence.

COMMUNICATION SKILLS

Communication skills, including speech, speechreading, manual communication, vocabulary development and other aspects of receptive and expressive language, of the Waardenburg subjects are similar to those of other deaf children. Approximately ten persons (50%) were considered to have above average language abilities; these are the same subjects mentioned previously who had above average IQs and tended to be leaders in their peer group. Those children with deaf relatives, especially deaf parents, tended to have better language. All of the ten persons with above average communication skills had deaf relatives. As might be expected, these skills were also better if the subject had more hearing; this would affect two of the ten persons mentioned.

Of the remaining ten subjects, nine were considered to have average communication abilities. The other subject had very poor language skills, which can be related to his educationally deprived background and poor home environment.

It is interesting to note that subjects imitated their backgrounds or models. Those children with an environmental situation involving motivation and deaf relatives who were leaders tended to follow in those paths. There were several cases where the subjects had better potential, but acheived only average communication skills, thus reflecting the attitudes of their environment and the language ability of their parents.

Consequently, it can be proposed that Waardenburg subjects will tend to have above average communication skills, especially if a motivational family background is present.

EDUCATIONAL ACHIEVEMENT

The average grade equivalent of six of the group who have graduated with known scores is 6.1. These do not include any of the exceptionally high or low achievers. This average is high when considering that thirty percent of the deaf population is functionally illiterate, and sixty percent achieve only a level of 5.3 or below (Mindel and Vernon, 1971). The known scores for those who have not graduated are given in Table 1, listing the subjects by chronological age and grade

Table 1. Educational Achievement of Waardenburg Subjects Still in School

Chronological Age	Grade Equivalent	Expected Grade Level*	Difference
17	5.1	5.7	- 0.6
14-6	6.9	5.7	+ 3.0
14-6	6.9	3.8	+ 3.1
13-11	5.9	3.8	+ 2.1
13-4	2.6	3.7	- 1.1
10	2.4	2.2	+ 0.2
6	1.8	1.5	+ 0.3

* based on "Academic Achievement Test Performance of Hearing Impaired Students," (1969)

equivalent at that age. The expected achievement levels and the differences between the actual and expected levels are also listed. All but

two of the subjects are functioning at least slightly higher than the predicted level. Of the seven subjects for whom scores were not available, teacher judgments indicated that four were classified as college material, one is still in school and well ahead of his class, one did well in school, and one did only average work, "average" being about fifth grade.

Educational achievement of these subjects can also be related to the individual's background, including education of the parents, motivation, family expectations, and cosmetic aspects. Many of these subjects may have achieved more if these influences had been more favorable. However, they are still achieving better than the "average" deaf student, and it can therefore be implied that persons with the Waardenburg syndrome will tend to have above average educational achievement.

SUMMARY

Because most attention has been focused on the physical characteristics of Waardenburg's syndrome, this paper has dealt primarily with the behavioral aspects. Retrospective research, using the existing school records of the Maryland School for the Deaf, was completed and indicated a prevalence of almost one percent of deaf children being affected by the syndrome. The subjects were well adjusted, with many having leadership qualities. Psychological adjustment seemed to be affected by the cosmetic manifestation of the syndrome, especially in the presence of cleft palate. Communication skills tended to be above average, with some evidence that linguistic skills were better developed in children from families where deafness occurred in several members. Intelligence averages in the bright normal range, and educational achievement is above the average of other deaf students. The environmental influences of the individual child have been noted. It seems apparent that while the Waardenburg syndrome has possible negative behavioral consequences, that these children tend to be highly educable.

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