CONGENITAL HEREDITARY VERTICAL NYSTAGMUS

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Case 1—A male aged 6 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 2—A female aged 15 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 3—A male aged 8 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 4—A male aged 10 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 5—A female aged 12 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 6—A male aged 14 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 7—A female aged 16 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 8—A male aged 18 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 9—A female aged 20 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 10—A male aged 22 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 11—A female aged 25 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 12—A male aged 28 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 13—A female aged 30 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 14—A male aged 32 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 15—A female aged 35 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 16—A male aged 38 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 17—A female aged 40 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 18—A male aged 42 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 19—A female aged 45 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 20—A male aged 48 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 21—A female aged 50 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 22—A male aged 52 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 23—A female aged 55 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 24—A male aged 58 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 25—A female aged 60 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 26—A male aged 62 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 27—A female aged 65 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 28—A male aged 67 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 29—A female aged 70 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 30—A male aged 72 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 31—A female aged 75 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 32—A male aged 78 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 33—A female aged 80 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 34—A male aged 82 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 35—A female aged 85 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 36—A male aged 88 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 37—A female aged 90 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 38—A male aged 92 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 39—A female aged 95 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.

Case 40—A male aged 97 years had shaking movements of the eyes and head, which were noted since birth. The condition is inherited.
stop them in a short time. The head movements disappear during sleep, but the nystagmus persists. It was observed that the head movements cease when the eyes are closed. The patient keeps his head habitually rotated to the right and the occiput slightly depressed towards the left shoulder, so that his eyes are directed somewhat to the left and upwards. The patient is left-handed and his Stanford-Binet I.Q. is 80. This patient was subjected to caloric tests as described by Fitzgerald and Hallpike (1942) in order to determine the function of the labyrinth, but there was no response to these tests.

Case 5 (IV 15).—In a female aged 15 months nystagmus was noticed at birth. The nystagmus is binocular, synchronous, and vertical in type, and the movements are pendular and of very wide range. Approximately 60 oscillations occur per minute. The rate is unaffected by fixing or looking to the side. The nystagmus is unaltered during sleep and there are no errors of refraction in either eye. Up to the present, no abnormal head movements have been observed.

Discussion

Nettleship drew attention to the fact that there seemed to be an association between the head movements and mode of descent in hereditary nystagmus. In some stocks the disease was transmitted by either sex, and affected females almost as frequently as males. This type was frequently associated with oscillation of the head. In another group the nystagmus was limited to males but was directly transmitted only by females and this suggested a sex-linked recessive factor. Oscillations of the head were rare or absent in this group. Glover (1937) described a family in which only males showed the defect and several had head nodding which tended to disappear. Rucker (1946) showed in his pedigree that the nystagmus was inherited as a sex-linked recessive factor and a considerable number of the affected persons had associated head movements. Duke-Elder (1949) subdivides congenital nystagmus into three groups:

(a) An ambly-sexual irregularly dominant form, affecting both sexes, but males more frequently than females; frequently there are associated head movements.

(b) A recessive sex-linked type, which is limited to males; head movements are not common, but defects in the eyes such as pigmentary failure, red-green colour blindness, and in the central nervous system, e.g., Lebers optic atrophy or spastic paralysis.

(c) A simple recessive rare form, which is frequently associated with consanguinity in the parentage. The pedigree described in this paper would be acceptable in the first group, except for the vertical nystagmus. It is not possible to draw any conclusions from such a small number of cases, but it would appear that the type of nystagmus is not tied to a particular mode of inheritance. Nettleship (1911), Cox (1936), and Walsh (1947) observed that the nystagmus was less marked during fixation and was increased by looking sideways. In this pedigree fixation and alteration of the visual axis had no effect on the nystagmus. It is interesting that some of these patients were unable to appreciate the nystagmus in others. It was thought at first that the eyes must be synchronizing, but in Cases 2 and 3 this does not seem possible as there was a considerable difference in the rate and amplitude of the nystagmus. Nettleship described a baby with rotary head movements, who died at 5 months of age. The sister, herself affected, was able to recall the head movements, but could not say whether or not the eyes were involved. It is possible that this girl could not appreciate the abnormal eye movements in her brother, and it is therefore important to obtain histories from unaffected members of the family, otherwise several cases may be missed.

Cox was of the opinion that the head movements cancelled the nystagmus in order to steady the retinal image, and refers to Nettleship as having made the same observation. This, however, is not necessarily correct, for according to Nettleship:

"So far as I have been able to observe, the two movements, when both are present, are in the same plane, both as a rule being horizontal. But whether the rapidity of the head-and-eye movements is the same and their rhythm such that one tends to neutralize the other I cannot say, and I doubt whether accurate information on the point can be gathered unless instantaneous photography or some kind of automatic movement-recorder can be pressed into the service."

In this pedigree and the case described by Posey (1902) the head movements were not in the same plane as the nystagmus, their rate was much slower, and the fact that the movements disappeared makes it difficult to imagine that the head movements were neutralizing or compensating for the nystagmus. M'Gillivray (1895) describes a patient with horizontal nystagmus and head movements, in which the head movements started with a nod, then rotation to the left, and this was followed by an upward movement with the result that the head movements roughly described a circle. In the other five cases the nystagmus was more rapid than the rotary head movements. M'Gillivray observed that the head movements were more active during fixation and Nettleship and Cox observed this in some of their cases. In this series the movements diminished during fixation and increased whilst gazing abstractedly. Case 2 volunteered the information that he was more conscious of the head movements while concentrating and he tried to suppress them as soon as they appeared.
The actual site of the lesion in congenital hereditary nystagmus is unknown as none of the cases have come to post-mortem study. Peripheral failure of fixation was suggested as an aetiological factor, but this could only apply if the nystagmus began after the first month, i.e., when the normal infant begins to fix. Nettleship (1911) describes cases where the nystagmus was present at birth and to this may be added Cases 4 and 5. Posey (1902) discussed his case with Professor W. G. Spiller (Philadelphia), who suggests that the vertical nystagmus might be due to weakness in the ocular muscles and that the lateral head movement might be due to slight left sterno-cleido-mastoid muscle weakness. Yawger (1917) referred his case to Dr. Alexander Bruce (Philadelphia), who suggested that Deiter’s nucleus might be the site of the lesion. Nettleship (1911) suggests that there may be a congenital lack of pigmentation of the retinal epithelium in the region of the macula. Walsh (1947) observed several negro families with hereditary nystagmus where pigmentation could scarcely be a factor. Hemmes (1926) thought that the nystagmus was due to a disturbance of the function of the otoliths. Duke-Elder (1949) thinks that anomalies may exist in the vestibular tracts and centres, or in the connexions between these centres and the oculo-motor nuclei, as well as a disturbance in the labyrinthine mechanism. It is impossible to say whether or not the labyrinth is normal in these cases, but Case 4 did not respond to the caloric tests. An unaffected member of the family gave a normal response.

**Summary**

Five cases of congenital hereditary nystagmus have been described. They are unusual in that the nystagmus was vertical in all, and in three patients the head movements were rotary in type. Linkage data are presented.

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**REFERENCES**


Williams and Wilkins, Baltimore.