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# EYE MOVEMENT RECORDINGS AS A DIAGNOSTIC TOOL IN A CASE OF CONGENITAL NYSTAGMUS*

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ABSTRACT

Objective recordings of eye movements in a patient with congenital pendular nystagmus showed only tiny superimposed saccades. Also, in tracking movements and in special "variable feedback" experiments, where a normal subject would ordinarily exhibit mainly fair-sized saccades, the patient showed only markedly decreased saccades. However, these small saccades showed maximal velocities appropriate to their reduced amplitudes; thus suggesting normal "time-optimal" motor control signal shapes and a normal extra-ocular muscular system. The increase in pursuit movements seems to be a compensatory effect secondary to the basic defect in saccades.

Prisms were prescribed to minimize the amplitude of the nystagmus since it had shown dependence upon angle of gaze and amount of vergence. Excellent subjective results were obtained and indicated that the sensory visual mechanism was normal.

Since the efferent signal shape, the muscular system, and the afferent visual system are all normal, and the smooth pursuit system better than normal (compensatory adjustment), we suggest that the basic defect in our patient is early in the generation of intermittent control signals for saccades from the central nervous system dual-mode controller.

The purpose of this study was two-fold—to analyze the nature of the control system disturbance in a well-documented case of congenital nystagmus and to utilize these findings toward providing a rational treatment for the patient. The patient was one of the authors of this article and had written his doctoral thesis on this topic[1].

The eye-movement recording methods and the objective findings are first described, next the clinical studies, treatment, and subjective results of treatment are given. Finally, in the discussion, certain pathophysiological features which could be identified are described.

## METHODS AND FINDINGS

The patient was especially cooperative and was aware of the nature of the studies and of the methods employed. These were the infrared reflecting eye movement recording glasses which are well described in the literature[2, 3, 4] and provide a comfortable and convenient method of recording eye movements of a wide variety of amplitudes. Indeed, as will be seen below, in order to display features of the eye movements over a wide range, it was not necessary to adjust recording conditions, but only to adjust the scale of the recording display.

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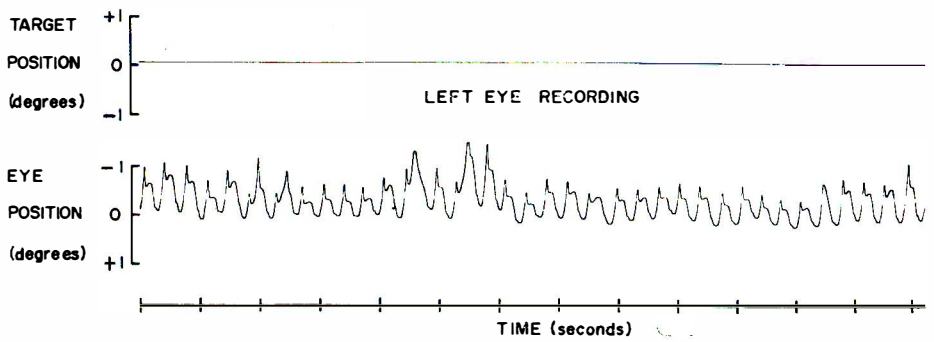


Fig. 1. High gain recording of eye movements during stationary target fixation; note basic smooth pursuit nature of nystagmus with a small superimposed saccade.

At high gain, as shown in Fig. 1, the nystagmus appeared to be a half to two-degree oscillatory movement occurring two or three times a second with quite definite component features. The main movement in each direction was a smooth-pursuit movement ranging from five to ten degrees per second in velocity. Superimposed on the extreme right position of the oscillation was a small saccadic jump to the right with an average amplitude of one-half degree. These could be clearly identified as saccades not only by their quantitative appearance on the recording, but also because their velocity of approximately 20 degrees per second fell along the characteristic nonlinear curve for velocity as a function of amplitude[5]; in this case the amplitude was one-half degree. We suggest that, in trying to define as saccades movements which range from small micro-saccades through corrective saccades to large saccades of tracking eye movements, those eye movements with a maximum velocity appropriate to nor-

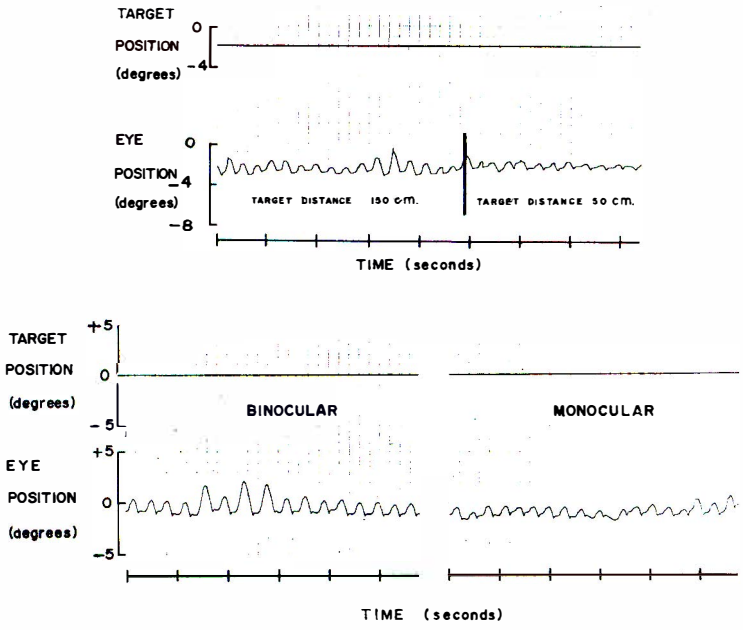


Fig. 2. *Upper:* Dependence of nystagmus amplitude on target distance and thus convergence. *Lower:* Nystagmus amplitude reduced in monocular fixation. Target distance 150 centimeters.

mal saccades be operationally defined as saccades.

The nystagmus in Fig. 1 could truly be called pendular nystagmus since the predominant movement is one of a smooth pursuit movement in both directions. The very small superimposed saccade suggests that there is some inability to generate large enough saccades to correct the nystagmus error such as is often seen in vestibular nystagmus, and also in optokinetic saccades. The patient showed a further number of individual characteristics as shown in Fig. 2: for example, the amplitude of the nystagmus was decreased when the subject viewed a target requiring a considerable amount of convergence effort. In addition, at times monocular vision also reduced the amplitude of the nystagmus—a finding for which we have no explanation.

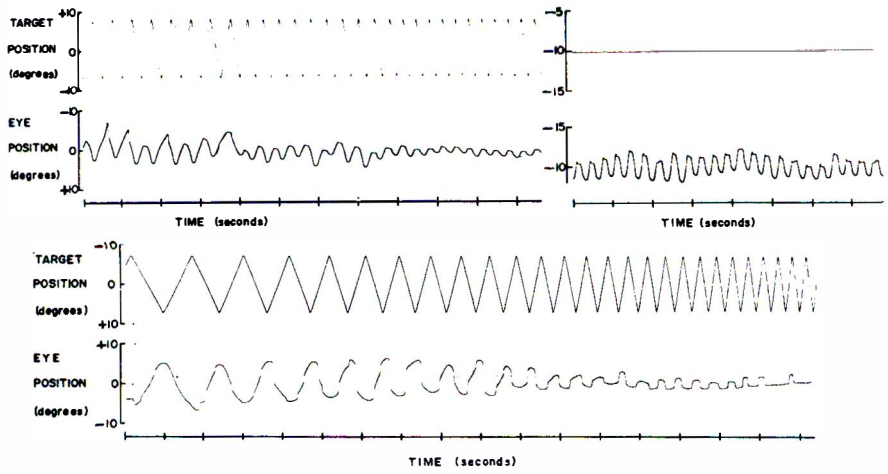


Fig. 3. Smooth pursuit tracking of target moving at constant velocity.
*Upper:* Shows the patient following to high frequency of alternation with low amplitude eye movements smaller than nystagmus movements on stationary gaze shown to right (different scale).
*Lower:* Shows normal subject unable to follow high frequency alternations and substituting saccades for smooth pursuit tracking.

We next studied the patient's ability to track target motions and noted a most interesting finding—that the patient could track more rapidly moving targets than the normal subject. In Fig. 3 in the upper left traces is shown the patient's response to an increasing frequency of alternation of constant velocity target motions. Here the scale is such that the very small amplitude saccades cannot be noted and the record consists almost entirely of smooth-pursuit movements. It can be seen that the patient has continued to keep up pursuit movements in phase with the target to quite high frequencies as compared with the normal subject in the lower traces. This normal subject record demonstrates the usual occurrence of both saccades and smooth-pursuit movements, then gradual elimination of smooth-pursuit movements at higher frequency, and finally the inability to track at all at very high frequencies. An interesting incidental finding is that when our patient was tracking there was some suppression in amplitude of his nystagmus: when the target was halted, as shown in the upper right traces of Fig. 3, the nystagmus became considerably larger than the tracking eye movements shown just to the left.

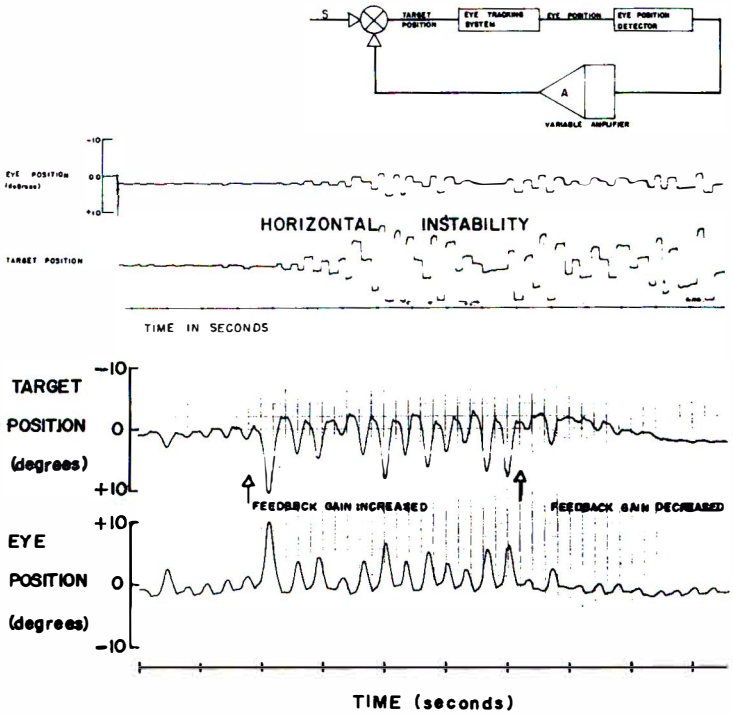


Fig. 4. A) Square wave oscillatory horizontal eye movements produced as an artificial instability by means of the variable feedback technique. Inset shows experimental arrangement with feedback from eye position detectors to target position stimulator via a variable amplifier. Redrawn from Gauthier Ph.D. thesis. Fig. 57, page 170.
B) High pendular nystagmus produced in patient by artificial variable feedback technique. Note pendular or smooth pursuit nature of these oscillations with dominant normal saccades.

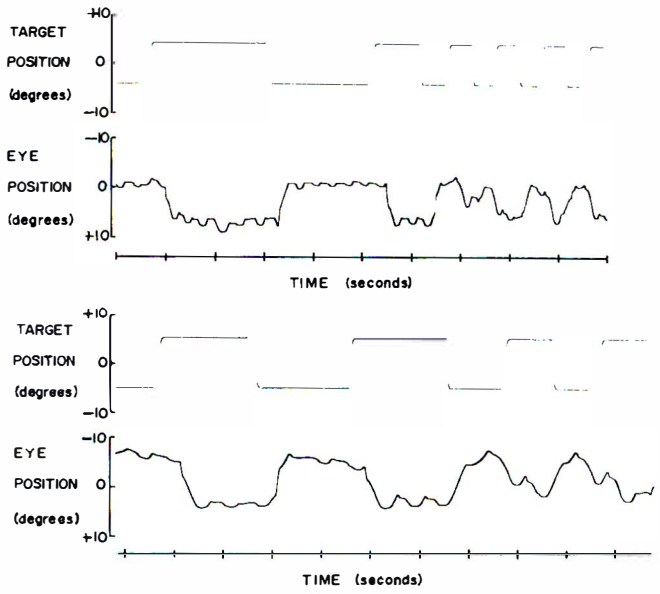


Fig. 5. Patient's tracking of a square wave target showing small inadequate saccades and the substitution of smooth pursuit movements to follow target. Also note nystagmus oscillations superimposed on tracking pattern.

When conditions of variable artificial feedback are produced[6, 7], a normal subject will break into a repetitive series of back and forth saccades which indicate a high gain instability of the saccadic eye-movement system as shown in Fig. 4A[8]. Our patient, as shown in Fig. 4B, developed high gain pendular nystagmoid movements under these conditions. Such movements might also be produced in the normal subject if, in addition to the high gain situation, the saccadic movements of the target had been removed either by linear filtering[9] or by nonlinear filtering[10]. Thus the patient's response would indicate the absence or attenuation of a saccadic system leaving his pursuit system to respond in this abnormal experimental situation.

In situations where saccades would be almost the only normal eye movement in normal subjects, the patient showed poor quality saccades, as well as superimposed pendular nystagmus as seen in Fig. 5. Here we see that the saccades, when they occur, are not sufficient to follow the stepwise target and are often continued by smooth-pursuit movements so that the subject is able to follow the target motion. The small component saccades seen with the patient's pendular nystagmus are also seen in the nystagmus superimposed on the tracking movements. The superimposed nystagmus was also influenced by the direction of the tracking movements and by the eye position as can be clearly seen in Fig. 6. Here the subject is tracking smooth ramps and sinusoids which provide opportunity for the nystagmus to be recorded in varying positions and during varying directions of tracking. It can be seen that the pendular nystagmus is of higher amplitude when the eye is rotated to a position toward the left; and also that markedly asymmetrical tracking occurs dependent upon the direction of the eye tracking motion.

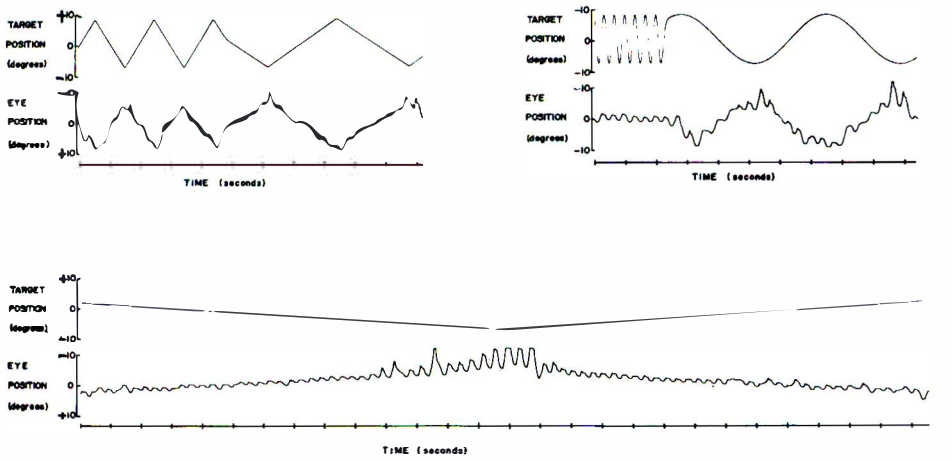


Fig. 6. Triangular and sine wave tracking: Shows nystagmus amplitude dependence upon eye position and tracking direction, especially well seen in lower traces with very slow wave target.

As mentioned above, the maximum velocity during saccades was measured, and our patient's saccades fell along the normal nonlinear saccadic velocity-amplitude relationship[5] as shown in Fig. 7 where the patient's saccades are plotted with triangles and are seen to cohere to the general nonlinear relationship.

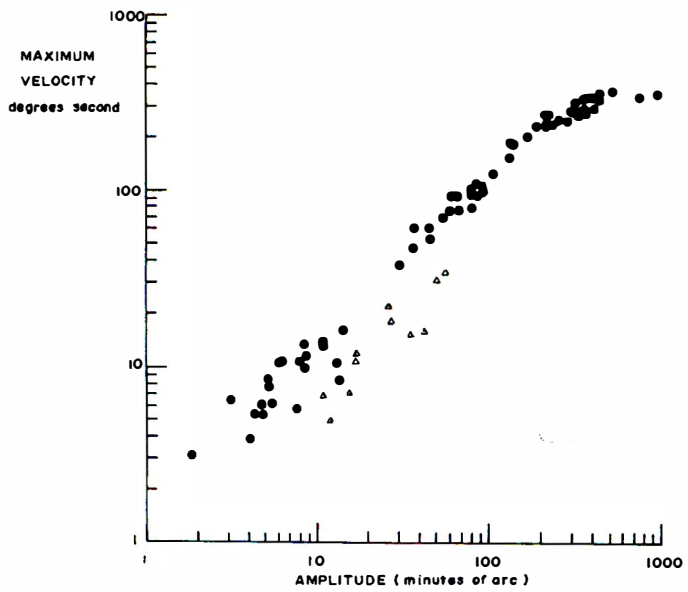


Fig. 7. Saccadic eye movement nonlinear velocity-amplitude characteristic redrawn from Zuber, et al.[5]

° for original data points and
Δ for patient's data points

CLINICAL STUDIES AND TREATMENT

Previous to our seeing the patient, he had been wearing the following spectacle correction:

O.D.: +2.00 D.S. ⌒ −2.50 D. C. axis 150
O.S.: +2.00 D.S. ⌒ −2.50 D. C. axis 15

This represented a full astigmatic correction and provided a binocular visual acuity of approximately 20/40. There were no prisms ground in the lenses. The patient's symptoms were eye fatigue and frequent intense headaches.

The pertinent clinical findings are as follows:

Subjective O.D.: +1.50 D.S. ⌒ −2.50 D. C. axis 150 20/30+
Refraction O.U.: 20/25−
O.S.: +2.00 D.S. ⌒ −2.75 D. C. axis 20 20/25−

The pendular nystagmus was grossly apparent. By external examination, the amplitude of the nystagmoid movements appeared to decrease with convergence. In normal straight ahead gaze, the patient habitually turned his head slightly to the right.

Changes of nystagmus with direction of gaze were shown in the previous section. Nystagmus amplitude was considerably decreased by lateral gaze of about two degrees to the left (Fig. 6)[a] and by convergence (Fig. 2). Lenses in spectacle form were designed to produce minimum nystagmus by directing the eyes to these corresponding positions (Fig. 8). Fig. 8 shows how versional prisms, base to the right, direct the eyes two degrees to the left to fixate a

[a]Reference 1 had previously quantitated this clinical finding. See, for instance, Fig. 2.11, page 49.

right. Fig. 8 shows base out vergence prisms producing convergence of the straight ahead target. This corresponds to eliminating the head turn to the eyes to fixate a straight ahead target. These two prismatic effects were combined in Fig. 8 (lower). Here the eyes converge a total of 16Δ and are rotated 4Δ to the left (1.73Δ = one degree). These composite prisms were incorporated into the astigmatic correction in the spectacle lenses. To aid the convergence necessary −1.00 D.S. were added to the spherical (hyperopic) correction.

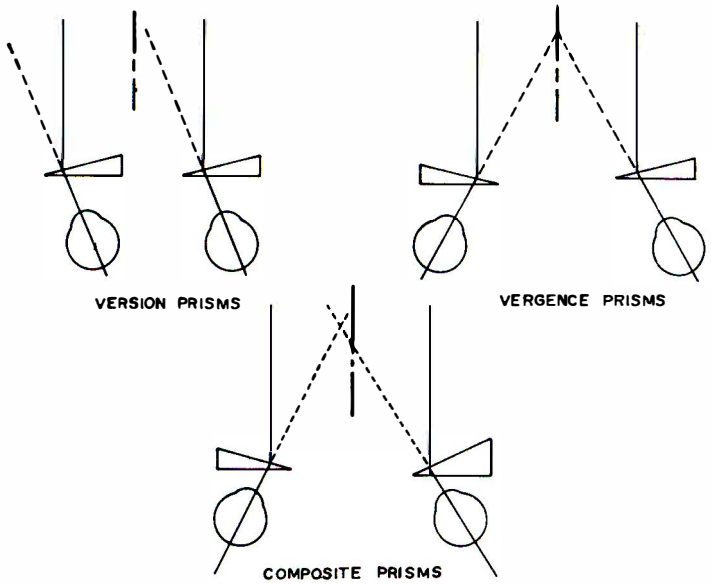


Fig. 8. Schematic representation of version, vergence and composite prisms.
*Upper left:* Version prisms—base to the right prisms which produce left version for straight ahead distant objects.
*Upper right:* Vergence prisms—base out prisms which produce convergence for straight ahead distant objects.
*Lower:* Composite prisms—base out and base to the right prisms which produce convergence and left version for straight ahead distant objects.

The final prescription was:

O.D.: +0.50 D.S. ⌒ −2.50 D. C. axis 150 ⌒ 10Δ B.O.
O.S.: +1.00 D.S. ⌒ −2.75 D. C. axis 20 ⌒ 6Δ B.O.

After two months of daily wear, the patient provided the following observations:

1. "Within one-half hour, new visual cues learned and uneasiness and nausea subsided."

2. "Vision very noticeably improved. I was able to see things which previously were not part of my perceived visual field (i.e., individual strands of hair on a person's head, TV scan lines, branches of trees and blades of grass), and while driving, I could read the license plates of other cars and road signs much more easily."

3. "My viewing distance for TV was increased due to the increase in visual acuity."

4. "Within 1-3 days the transition period associated with removal of

glasses or putting them on was reduced to one second."

5. "Eyes much less tired at day's end."

6. "Headaches, which used to be common, drastically reduced in frequency and intensity."

The family tree of our patient is seen in Fig. 9 (lower). The children of the marriage of first cousins and their descendants are shown in detail. Only males are affected and these are descendants of half the daughters of the previous generation; this is characteristic of a sex-linked recessive.

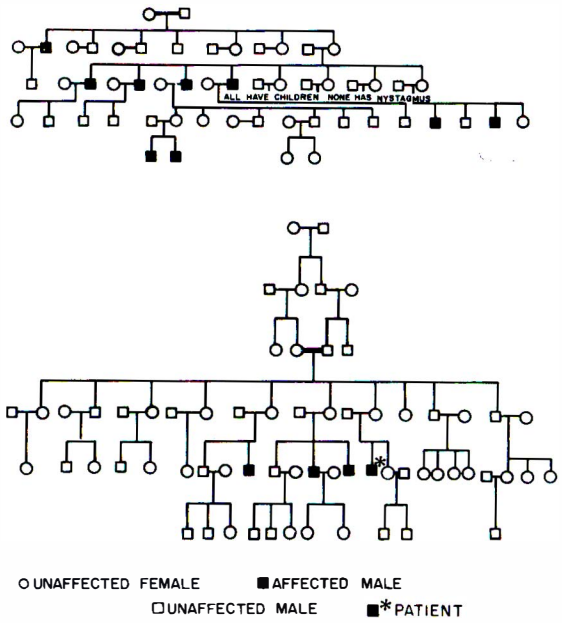


Fig. 9. *Upper:* Nystagmus through four generations showing discontinuous inheritance with only males affected. Redrawn from Billings, 1942.
*Lower:* Family tree of our patient showing the first occurrence of nystagmus in the grandsons of the marriage of first cousins. Note that in each case the occurrence in sons of daughters of the marriage of first cousins.

## DISCUSSION

The use of the maximum velocity of an eye movement to derive an operational definition of a saccade seemed to us to be a useful route to follow. Saccades can be defined by a number of their features, such as the intermittency[6, 7], the saccadic suppression[11, 12, 13] that accompanies them, and by their dynamical shape[3, 4, 14]. This latter finding is the easiest to record and ascertain in the case of patients where extensive experiments may not be possible.

Pendular nystagmus[15] can be secondarily defined as nystagmus which does not have a saccade as a consistent and major feature of its dynamics[16, 17, 18], even though saccades may exist to some extent concurrently. Our patient is an example of this where the saccades seemed to occur with most of the nystagmus movements but had very little effect on the eye movement position since it was small and, indeed, hard to see on a display scale that showed any type of other movement, such as when the nystagmus was superimposed on an eye-tracking movement.

One of the main findings following our successful treatment of the patient was that his central vision was quite accurate. This enables us to conclude that defect in sensory vision is not the primary pathophysiological mechanism[15]. We must therefore search into the central nervous system control mechanisms for the pathophysiological origin of the nystagmus before proceeding with our somewhat speculative attempt at this analysis. We would like to point out that with a long-standing difficulty, such as the pendular nystagmus in our adult subject, it is quite possible that many of the findings might be as a result of compensatory mechanisms rather than due to the basic defect.

The control system for eye movements has at least three major features: the predictive mechanism, the ocular-motor dynamics, and the dual mode controller. The predictive mechanism[2] which enables a human subject, but not a monkey[19], to predict target position in the case of simple target motion, such as triangular and sinusoidal oscillations. These responses do not appear to be basically interfered with in our patient. We have not tested this extensively; but for example, the subject seemed to predict repetitive square waves and started his regular pattern at times in synchrony with the regular target motion. Similarly, the ocular-motor or plant dynamics seemed to be within the normal range. His smooth-pursuit movements could reach reasonably high velocities, certainly within normal limits. Most important, as mentioned above, his saccadic movements, although small, infrequent and often strikingly absent, when they occurred seemed to have dynamics that fell within the normal range[5] as shown in Fig. 7. This would indicate that the pattern of activation, force generation, apparent internal viscosity (force-velocity relation), agonist-antagonist, reciprocal innervation, passive orbital and muscular loading were all normal[3]. The possibility of decreased force generation, or of a consistent increased elasticity cannot, of course, be completely ruled out as contributing to the pathology, but seems less likely to us at the present time, especially since smooth-pursuit movements were so well performed as in Fig. 3.

We, therefore, would tend to put the pathogenetic processes at some point within the dual mode controller[19] which is generating the control signals for saccades. If this were pathological, we would then expect to find as we did that saccades were frequently absent on tracking squarewave target motions, and when saccades occurred, they would be small and incomplete.

The more active use of the smooth-pursuit mode might develop as a compensatory mechanism. This increased reliance on pursuit movement might in turn produce defects secondary to compensatory action. For example, if the gain of the pursuit system were turned up, instability might arise and make its appearance in the form of pendular nystagmus. The increased pursuit movements might sometimes even be more effective than in the normal subject, such as when tracking fairly rapid ramp targets as shown in Fig. 3. However, again we would like to emphasize that the increased pursuit movement may not be the primary defect, but may be compensatory to the absence and attenuation of saccades.

The effect of both direction of gaze and the amount of vergence on the amplitude of the nystagmus would tend to support the idea that an abnormally active pursuit movement system could be related to the present nystagmus

since these maneuvers would tend to change the tonic level of innervation. Indeed, our prescription exploited both of these conditions in an attempt to control the nystagmus!

Many of these ideas should be considered as speculative hypotheses and will merely serve us as a base for further detailed study of other patients. We are also planning to model the dual-mode control system with the indicated abnormalities to see if we can obtain the qualitative features of our patient's nystagmus as shown in Figs. 1 through 6 and the quantitative findings in Fig. 7.

Forssman[20] had reported on 90 cases of congenital idiopathic nystagmus utilizing EOG method of measuring eye movements. He quotes Hermes in estimating the incidence at 1:6500. He reports that the genesis is unknown and suggests it to be of hereditary nature. He further suggests the formal cause of many cases must be in the CNS since the nystagmus was present the first day after birth. Forssman refers to Kettleship (1911)[20] and Kaser (1942)[20] as describing the decrease in nystagmus when the patient fixates a near target. Gamble (1934)[20] is quoted to have found in 130 eyes 118 with astigmatic refractive errors. Astigmatism and nystagmus has also been frequently reported in patients with ocular defects, such as albinism[21].

Waggoner and Boyd[22] have reported nystagmus in one family followed through four generations. The inheritance is discontinuous affecting males only. Five of the seven males in the fourth generation were affected. They conclude sex-linked inheritance and point to consanguinity as an important etiologic factor. Billings[23] reported a similar case through four generations (see Fig. 9, upper). She also concludes that the mode of inheritance is as a sex-linked character.

Thus our clinical study shows our patient to be typical of previously reported cases of this "nonsensory pendular nystagmus." The results of our clinical treatment are noteworthy because of our use of composite prisms, also reported by Metzger[24], and because of the reliability of the assessment of subjective results by our Ph.D. patient.

The absence of defect in the ocular-motor "plant" dynamics as demonstrated by our quantitive eye movement recording studies is important especially when taken together with the absence of sensory defect. Thus localization is possible to a pathophysiological defect in saccadic movement generation with a compensatory reliance on the smooth pursuit system and possibly a further generation of instability in this latter system to generate the pendular nystagmus itself.

SUMMARY

1. Objective recordings of eye movements from a case of congenital nystagmus are presented.

2. Patient showed decreased saccades in conditions where one would normally expect them and in abnormal feedback conditions where a normal subject would show them. The nystagmus was of a pendular type with only very small superimposed saccadic movements. These small saccades fell within the normal range of maximum velocities for their amplitude thus suggesting a normal orbital muscular system. These have a number of other quantitative

findings, suggesting that the abnormality is located in the dual mode control signal generator with a deficit of saccades and a compensatory increase in pursuit movements. However, it is difficult to distinguish between a basic defect and a compensatory movement in this adult patient.

3. The amplitude of nystagmus showed depends on angle of gaze and amount of vergence. Prisms were prescribed to minimize the amplitude of the nystagmus.

4. The subjective results were excellent and thus indicated that a visual defect was not the primary one.

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