DISEASE WITH ABNORMAL EYE MOVEMENTS

5.1 In normal subjects, the eye movement response with head fixed consists of a superimposed response of the saccadic and smooth pursuit systems (1). These two systems have a common effector and receptor viz. extra-ocular muscles and the orbit. The eye movements exhibit a high degree of variability (2) and hence their central control actions are difficult to analyse. More knowledge about the mechanisms controlling these systems may be obtained if experiments are done on subjects whose two types of eye movements are differentially affected due to disorders at the higher centres of control. For example, if experiments are conducted on subjects whose saccadic movements are damaged or lost, due to a disorder at a higher center of control, then simple step and ramp-step responses of eye movements of these patients will reveal valuable data about the dynamics of the smooth pursuit system. In such a case, complicated experimental techniques such as use of electronic filters to differentially affect the feedback factors for saccadic and smooth pursuit movements (Chapter V), designing special ramp-step stimuli to elicit only the smooth pursuit response or pharmacological intervention to eliminate one of the movements (Chapter III), may be avoided.

5.2 A group of patients suffering from a form of "heredofamilial Spinocerebellar Degenerations" were found to exhibit
unusual eye movements (3). The eye movements of these patients were studied in detail. The results of experiments done are described in Chapter VI.

5.2.1 Family and Ethnic Background

About 30 case histories of patients from 9 families suffering from this disease are known. The disease was strongly familial affecting several generations in six families, while the other 3 were sporadic cases. There was no parental consanguinity in any family (Fig. 5.1). Seven families were Hindus and two Muslims from Maharashtra. Two families had moved from neighbouring Gujarat and one from Sindh (Pakistan) for over 20 years. The inheritance appears to be a non-sex-linked autosomal dominant gene. The disease affected 19 males and 11 females. Age of the onset of the disease could be established in 22 patients. The average was 22 years, the oldest being 30 years and the youngest 8 years.

A representative case history of one of the patients is as follows:

The propositus, who was 11 years old, had difficulty in walking, disability in the use of his hands and tremors from the age of 8 years. A year later, speech started becoming incomprehensible. He was an intelligent child. A moderate cerebellar ataxia of the limbs and trunk and dysarthria were noted. The deep reflexes were sluggish. The plantars were flexor. There was no history of diplopia,
FIG. 5.1 FAMILY TREES OF THE PATIENTS

PEDIGREE OF APT FAMILY

APR - SUBJECT "C"

(b) AB family

LEGEND

History of Similar Illness

Relatives Examined - NORMAL

Dead

Male

Female

PATIENTS

(c) PPP - Subject "E"
Fig 5-1 (d) PEDIGREE OF PSJ FAMILY
SUBJECT F

(e) PEDIGREE OF GH FAMILY
SUBJECT G
ptosis or squint. There was no nystagmus. There was no limitation of ocular movements; but random and command movements were slow.

After four years, the patient was worse and could hardly walk. He was mentally retarded, the ataxia was marked, and there was constant intention tremor of the whole body. The eye movements were much slower and accompanied by head jerking which had been very slight previously. There was a slow sweep of eyes when trying to look at objects. No disconjugate deviation of the eyes was noticed, and the pupils were normal. The patient constantly jerked his hand as he scanned his surroundings and often blinked his eyes. There was some weakness and slight wasting of the facial musculature. Choreiform movements of the face and hands were present. There was wasting of the soles of both feet, and the deep reflexes were absent. The plantars remained flexor.

5.2.2 Clinical Investigations

1) Routine investigations (B.P., Urine etc.) including CSF examination, were normal.

2) Caloric stimulation: The ears were irrigated by cold (30° C) and warm (44° C) water in 12 patients. In 9 of them, no nystagmus was elicited. On irrigating with cold water, the eyes deviated to one side and with warm water to the opposite side. The return to normal position occurred on an average from 1 minute and 35 seconds to 2 minutes and 40 seconds after stopping irrigation. In 2 patients (milder cases) the eyes tended to deviate into the corners, but did not get
fixed there as in others, but came back to mid-line with jerky movements. In the remaining patient, slow oscillations of the eyes replaced the expected nystagmus on irrigation with cold and warm water.

3) Ba'ra'ny's rotation test - This test was performed on 12 patients. Each patient was rotated in a Ba'ra'ny's chair ten times in 20 seconds. The eyes deviated to the opposite side at the beginning of the rotation (on clockwise rotation, eyes deviated to left) and remained there till the rotation was stopped. On stopping, it was noticed that the eyes swung in the direction of rotation. The eyes came to the mid-line gaze, after repeated blinking or head jerking within from one minute 30 seconds to 2 minutes and 40 seconds. This was found in 10 of the 12 patients on whom the test was performed. One patient showed deviation of the eyes as in others, but the eyes returned to the mid-line with small nystagmus without any head jerking or blinking. The remaining patient also showed the same initial deviation, but on stopping the rotation, showed oscillatory movements of the eyes.

4) Optokinetic Nystagmus. An attempt to obtain an ocular response to a rotating drum marked with black and white strips was made in 13 patients. The expected response was absent in all of them. No distinct nystagmus was obtained, except in the vertical direction in one mildly affected patient. In 2 others (less affected), side to side slow oscillations were seen, when the drum was moved slowly but disappeared on increasing the speed. In others, no nystagmus could be
demonstrated even after variation of the speed of rotation.

5) **Electromyography** It was performed in 11 patients out of which 9 showed patterns of chronic denervation of the facial muscles and of the limb (especially of the lower limb) muscles. Medial and lateral recti of one or both eyes were examined in 7 patients and were found to be normal every time.

6) **Pneumoencephalography** It was performed in 11 patients and was found normal in only 2 of them. In the others, a large cisterna magna with a fluid level was visualized. In some of them, enlargement of the fourth ventricle was also seen. All these 9 patients had mild to moderate enlargement of lateral ventricles and with excess of air in the subarchnoid space over the frontal lobes.

7) **Muscle Biopsy** The quadriceps muscle was examined in 4 patients and in 3 there was evidence of "neurogenic atrophy". The remaining one was normal. The lateral and medral recti of the eye were normal in all 3 in whom they were examined.

8) **Endrophonium** - There was no improvement in the ocular movements after administration of endrophonium 10 mg intravenously in any patient.

9) **Cerebellar** - The disease began in all patients with imbalane and difficulty in walking and later proceeded to affect the upper limbs and the speech. Trunk ataxia was often
severe. Postural titubation and intention tremors were seen in the more severe cases. Most of the patients' speech was incomprehensible.

10) Oculomotor - All 16 patients had abnormal ocular movements and the severity varied with the severity of the disease. None had ocular or visual symptoms; No squint was noticed although sometimes a slight loss of parallality in either eye briefly became evident. Convergence was easier than other movements. There was no ptosis or pupillary abnormality. Two principle signs were (1) slowing of eye movements, (2) abnormal accompanying movements of the head and neck which were directly related to the severity of the former. Whether the eyes moved randomly, on command or on following objects, the movements were slow. The quick, random scanning movements of the eyes seen in a normal person were replaced by a staring look. Most of the scanning of the surrounding was done by head movements accompanied by a slow drift of the eyes in that direction. On command to look in one direction with the head fixed the eyes moved slowly although fully. There was no limitation of the ocular movements in any of the patients, although, in the more severely affected more than one attempt had to be made by the patient to move the eyes into the corners. This was easier if the head was left free because the eyes moved more easily with head jerking. Occasionally blinking helped to 'unfix' the eyes. Vertical movements were easier than horizontal and downward gaze was the easiest. With head fixed, reading and writing became difficult.
Fig. (5.2) shows other physical signs. Among these unusual features were the weakness of the facial masculature in 8 patients and choreiform movements in 4. The wasting was not severe but it gave rise to hollowing of the cheeks and temples. The choreiform movements, confined mostly to the face and hands were usually mild whereas the cerebellar and oculomotor signs were seen in all the members examined, the other features were randomly scattered through the various families e.g. extensor plantar responses were seen in patients with both absent and exaggerated deep reflexes.

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