

Eye Movoment Changes in Albinism

— A Case Report with Electronystagmographic Findings —

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Albinism is associated with neural anomalies including foveal hypoplasia and aberrant optic pathway projection that result in a variety of oculomotor instability. We present a 38-years-old man with oculocutaneous albinism who had horizontal jerk-type nystagmus, which showed a reverse in direction by any extraneous light stimulation which was documented by electronystagmogram. The mechanism of the nystagmus in this case is uncertain. These findings in albinism have not been reported previously, to our knowledge, and suggest a defect in the visual pathway system.

Key Words: Albinism, eye movements, electronystagmogram

Albinism, a genetic defect preventing normal synthesis of melanin, is accompanied by profound disturbances of the visual system, such as low visual acuity, photophobia, strabismus, and abnormal eye movements (Apkarian *et al.* 1983; Kinner *et al.* 1985).

The most likely causes of underlying abnormal eye movements are foveal abnormalities which result in degraded sensorimotor feedback, and optic pathway anomalies resulting in directional errors of sensorimotor feedback (Collewijn *et al.* 1985). Abnormal eye movements have been more directly associated with the neuroanatomical abnormalities of the optic pathway, but not without controversy (Collewijn *et al.* 1985). A large proportion of optic nerve fibers originating in the temporal retina, which normally do not decussate, are misrouted and terminated contralaterally (Guillery *et al.* 1975). The misrouted optic pathway has been

documented anatomically (Guillery *et al.* 1975) and electrophysiologically (Creel *et al.* 1978; Guo *et al.* 1989; Russul-Eggitt *et al.* 1990). We report here for the first time, as far as we know, an albino patient with nystagmus reversed in direction by any extraneous light stimulation which was confirmed by electronystagmogram.

CASE REPORT

A 38-year-old, right-handed man with oculocutaneous albinism was referred to the department of Neurology, Young-Dong Severance Hospital, because of dizziness and headache. There was no known family history of genetic illness. His birth and development were not remarkable. From early age, white hair, white skin with a tendency to sunburn very easily, photophobia, abnormal eye movement and poor vision were noted.

On examination, the patient appeared photophobic, and had dry, pinkish white skin and white hair. There was an obvious horizontal jerk-type nystagmus. He had congenital esotropia of the right eye. His visual acuity was 10/200 in each eye. The pupils were

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isocoric and reactive to light and the irises were hypopigmented. The fundus was observed to be hypopigmented with easily visible choroidal circulation (Fig. 1). Other neurological examination including cranial nerves, cerebellar testing, motor system, and sensation were nor-

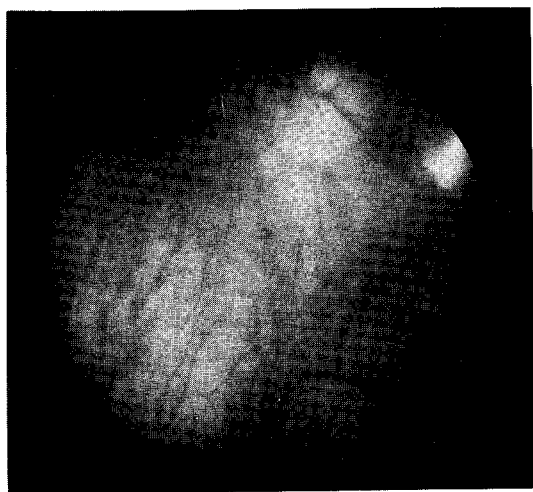


Fig. 1. Fundus showed hypopigmented with easily visible choroidal circulation.

mal. Laboratory tests including CBC, urinalysis, serum electrolyte, ESR, EKG, liver function test, and renal function test were normal. Pure tone audiogram was normal and stapedial reflexes were intact. Electrodiagnostic studies showed normal nerve conduction velocity (NCV), brainstem auditory evoked potentials, and posterior tibial nerve stimulation somatosensory evoked potentials. However, visual evoked potentials using pattern shift stimulation revealed no cortical waves bilaterally. Radiological studies including chest PA and brain CT scan were normal. A detailed analysis of the patient's nystagmus was undertaken with electronystagmogram.

Electronystagmographic findings

We tested spontaneous nystagmus under different viewing conditions (light and darkness), optokinetic nystagmus (OKN), optokinetic after nystagmus (OKAN), bithermal caloric test, as well as smooth pursuit and saccadic eye movements (Table 1).

The spontaneous nystagmus was observed. Horizontal jerk type nystagmus, which was reversed in direction, was left beating in the dark room and right beating in any extraneous light (Fig. 2). The optokinetic visual stimulation

Table 1. Electronystagmographic findings in albino patient

Examination		Findings
1) Spontaneous Nystagmus	in any extraneous light in dark	Rt beating Lt beating
2) Optokinetic Nystagmus	leftward stimulation 40 deg/s leftward stimulation 10 deg/s leftward stimulation 40 deg/s leftward stimulation 10 deg/s	Rt beating Rt beating Rt beating None
3) Optokinetic after Nystagmus	leftward stimulation 40/deg/s rightward stimulation	Rt-Lt beating Lt beating
4) Smooth pursuit	Stimulation velocity 5 deg/s 20 deg/s 40 deg/s	Gain=0.82 0.36 0.07
5) Saccadic	Random 6~32 degree/sec jumps	Impaired
6) Bithermal caloric	left ear, cool right ear, cool left ear, warm right ear, warm	Lt-Rt beating Lt beating Lt-Rt-Lt beating Lt-Rt beating

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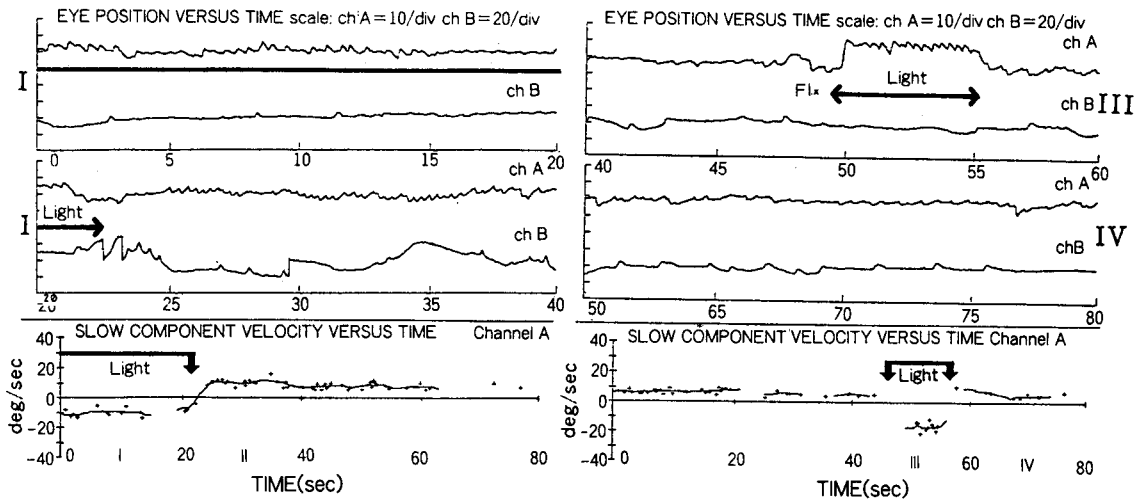


Fig. 2. Records of spontaneous nystagmus showed changes in direction under conditions in the dark and light. Ch A; horizontal eye channel, Ch B; vertical eye channel.

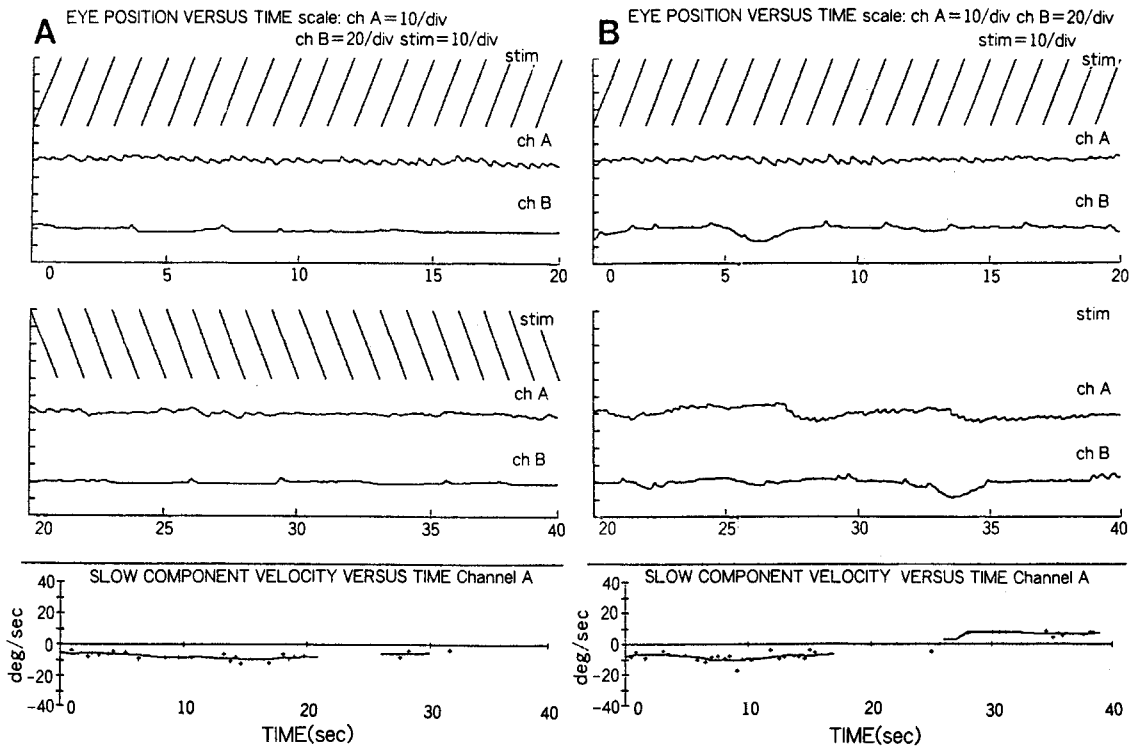


Fig. 3A. Records of optokinetic nystagmus, Stimulation; Horizontal bi-direction 40 degree/second, Ch A; horizontal eye channel, Ch B; vertical eye channel. Upper trace shows optokinetic nystagmus with leftward stimulation and lower trace shows optokinetic nystagmus with rightward stimulation.

Fig. 3B. Records of optokinetic after nystagmus, Stimulation: Horizontal leftward, 40 degree/second, Ch A; horizontal eye channel, Ch B; vertical eye channel. Upper trace shows optokinetic stimulation with leftward stimulation, and lower trace shows optokinetic after nystagmus without stimulation.

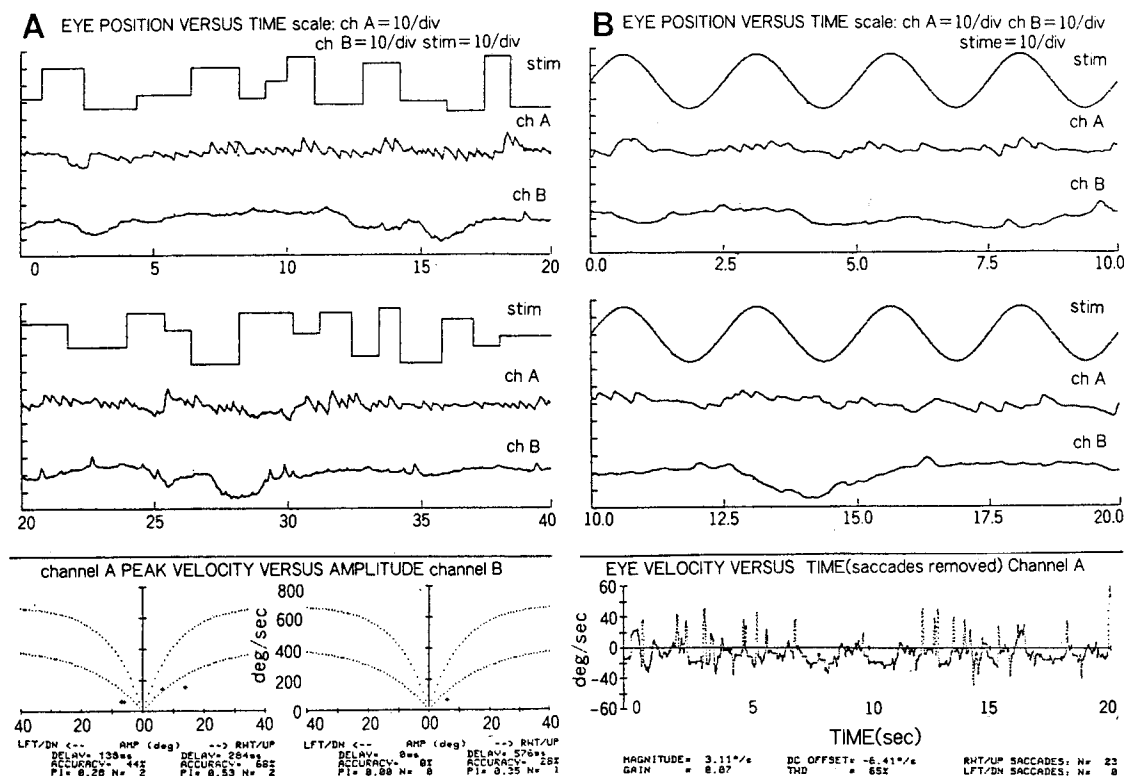


Fig. 4A. Records of saccadic eye movements; Stimulation: random 6~32 degree/sec jumps, Ch A; horizontal eye channel, Ch B; vertical eye channel.

Fig. 4B. Records of smooth pursuit eye movements; Stimulation velocity=40 degree/second, Ch A; horizontal eye channel, Ch B; vertical eye channel.

drifted at a fixed velocity, 40 degree/second and 10 degree/second, with a target spacing of 2.56 degree, from right to left or left to right. The patient viewed the light target binocularly from a distance of 91 cm. The optokinetic nystagmus was not seen during optokinetic stimulation, but there was only a spontaneous right beating nystagmus (Fig. 3A). We also observed optokinetic after nystagmus for 20 seconds in the dark room after 60 seconds optokinetic stimuli. The direction of optokinetic after nystagmus was right beating during the initial 5 seconds and followed by left beating after leftward stimuli (Fig. 3B), and left beating nystagmus after rightward stimuli. Examination of visual tracking of a light dot moving at speeds of 8, 20 and 40 degree/s showed an impaired state and there was a slightly right beating nystagmus (Fig. 4). The severity of the degradation

in smooth pursuit increased dramatically with an increase in stimulus velocity. Examination of horizontal random saccade with 6-32 degree jumps also showed a severely impaired state (Fig. 4).

After bithermal caloric stimulation in the sitting position with the head tilted 60 degrees backward, eye movements were recorded in the dark room. Infusion of cool water (30°C) into the left ear for 30 seconds induced a left beating nystagmus in the early period and delayed the appearance of the right beating nystagmus later. Infusion of cool water into the right ear induced a left beating nystagmus. Infusion of warm water (44°C) into the left ear induced a left beating nystagmus in the early period, and then right beating later, and thereafter the left beating nystagmus reappeared. Infusion of warm water into the right ear induced a left

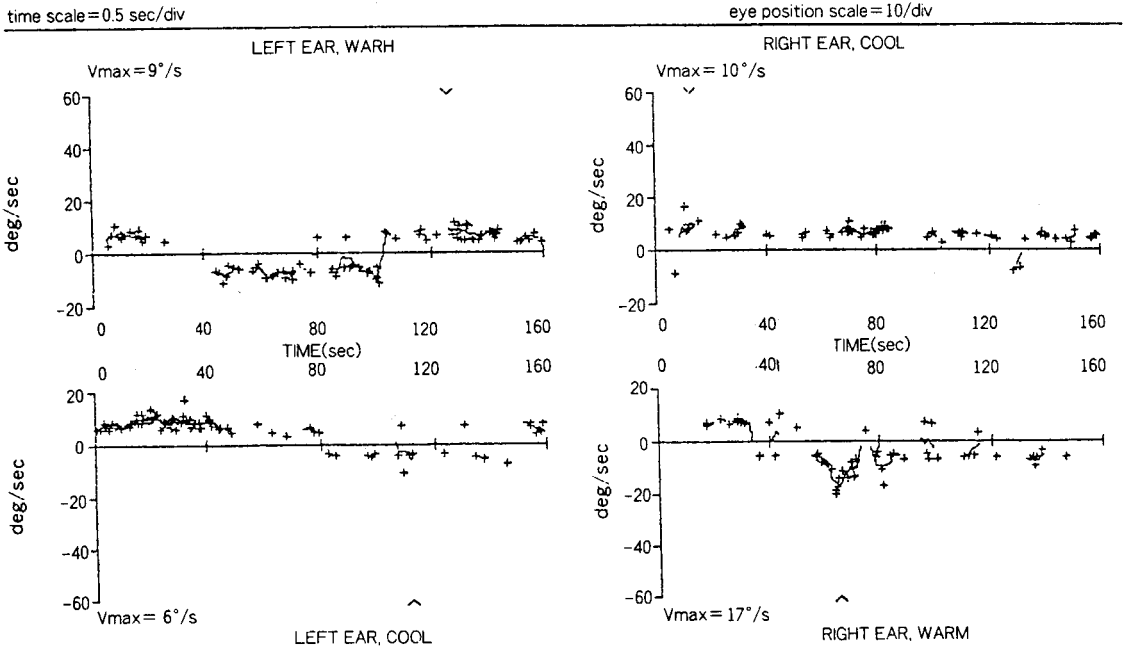


Fig. 5. Records of bithermal caloric test; slow component velocity versus time chnnel A.

beating nystagmus in the early period and delayed right beating nystagmus (Fig. 5).

DISCUSSION

The visual system anomalies in albinism are foveal hypoplasia and aberrant optic pathway projection, which result in two major visual complaints: reduced visual acuity and abnormal oculomotor instability. The oculomotor instability shows a variety of eye movement disturbances, with considerable intersubject variability. These individuals may have pendular or jerk nystagmus, absent or inverted optokinetic nystagmus, inverted or defective pursuit when targets are projected onto the temporal half-retina (Collewijn *et al.* 1978; Collewijn *et al.* 1985; Halmagyi *et al.* 1980; Simon *et al.* 1984). Periodic alternative nystagmus may also occur (Davis and Smith 1971; Guyer and Lessell 1986). The nystagmus often damps when closed vision is being used and, conversely, becomes more marked in bright illumination, with a resultant reduction in vision (Krill 1977). The intensity of

nystagmus often lessens with age, and this may be associated with improvement in visual performance (Krill 1977).

In our albino patient, we could observe an unusual nystagmus that reversed in direction. The nystagmus was always left beating in the dark room, turning to a right beating nystagmus in a lighted situation (light on or any visual stimuli), and turning again to left beating immediately after the room light had been extinguished (Fig. 2). We could also observe a right beating nystagmus even during the pursuit and saccadic stimuli. The optokinetic nystagmus may be impaired because there was only a spontaneous right beating nystagmus which may be a spontaneous nystgmus by light stimuli. The optokinetic after nystagmus may be normal response. Bithermal caloric responses were seen but were abnormal. The left beating nystagmus in the early period after caloric stimuli was probably produced by the elimination of light after caloric stimulation in the light room and the following direction changed nystagmus is due to the response of caloric stimuli on the peripheral vestibular organ. These responses suggest a normal state of the peripheral vestib-

ular organ. John RS *et al.* (1984) described five albinism patients, they found the four of the five patient showed changes in nystagmus under conditions of light and darkness. One patient showed a suppression of nystagmus in the dark. Three patients showed an increase in the amplitude of nystagmus in the dark. Collewijn *et al.* (1985) also described that nystagmus reversed in direction by responding to targets. To our knowledge, it had not been reported in an albino patient that nystagmus reversed in direction by any extraneous light stimulation.

One of most reliable sign of albinism is asymmetry of visual evoked potential, however, our patient had no visual evoked potential using pattern shift stimulation. We thought that this finding was probably due to the patient's poor visual fixation because of poor acuity and severe photophobia although we could not obtain visual evoked potential using flash stimulation.

How might albinism cause nystagmus? The nystagmus may be caused by the subnormal visual acuity and by the aberrant visual pathway projection. Boylan and Harding (1983) have suggested that the nystagmus seen in albinos might be the result of poor central fixation from lack of foveal differentiation, and Fulton *et al.* (1978) have demonstrated that the fovea is absent in albinos. However, Collewijn *et al.* (1985) have proposed evidence showing that poor acuity does not cause the nystagmus in human albinos. They point out that albinos first manifest nystagmus at an age when normally lacking foveal differentiation and also that while all albinos lack foveal differentiation, some do not have nystagmus.

The aberrant visual pathway in albinos is well documented anatomically and functionally in the literature (Creel *et al.* 1978; Drager 1986; Guillery *et al.* 1975; Guo *et al.* 1989; Russel-Eggitt *et al.* 1990). Although the view is still controversial, most authors believe that aberrant optic pathway projection is the primary cause of the oculomotor instability (Collewijn *et al.* 1985). The aberrant crossing fibers are connected to the wrong hemisphere and give rise to disorganized cortical maps. The anomalous inflow could potentially generate oculomotor instability.

In our patient, the aberrant optic pathway may be associated with unusual oculomotor behaviors, but the mechanism is uncertain.

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