

Short report

Familial congenital vestibular areflexia

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SUMMARY Three cases in one family are presented with oscillopsia due to vestibular areflexia, but without hearing loss. There was no history of other neurological or otological diseases (including infectious diseases) or use of neuro-ototoxic drugs. Laboratory tests, including tests for autoimmune diseases, were undisturbed. Petrosal radiographs and brain CT scans were normal. The pedigree suggested autosomal recessive inheritance.

Some patients report the symptom of illusory movements of the visual world during head movements only, which is known as head movement-dependent oscillopsia. The illusory movements take place along the same axis as the head movements. This type of oscillopsia is found for example in patients with loss of function of the vestibular end organ.¹⁻⁸ We present three cases in one family with probable congenital vestibular areflexia.

Cases

A 37 year old man (fig, F) complained of blurred vision and illusory movements of the visual world during head movements. Walking was broad-based and very difficult in the dark. Reading during head movements was almost impossible. When swimming underwater, loss of spatial orientation was experienced. As far as he remembered, the patient recognized these complaints at school age. He worked on a ship for 5 years and was never sea-sick. His 52 year old sister (fig, D) mentioned difficulties during walking in the dark from the time she was a little girl. One day, when she was swimming underwater she experienced also loss of spatial orientation. The 55 year old brother (fig, C) of these patients also noticed illusory movements of the visual world and difficulty when walking in the dark as long as he could remember. He worked at sea for 12 years but never experienced sea-sickness. All three patients had difficulties in looking around while riding a bicycle; they tended to fall. Another brother (fig, E) had drowned at the age of 26 years;

after a night party he got into the water, while he was attempting to get on board his ship. Their history was negative for central nervous system and otologic infectious disease or the use of neuro-ototoxic drugs. Their father (fig, A) never had similar complaints. He was a good swimmer and diver. He died of cardiovascular disease at the age of 63 years. Their mother (fig, B) had no complaints. Other members of the family (see pedigree fig 1) and including the three brothers and four sisters of (A) and three sisters and five brothers of (B) as well as their children gave no history of a similar complaint. Physical and neurological examination was normal except for oscillopsia starting at a frequency of roughly 1.5 Hz passive head movements (amplitude roughly 20°) in cases C, D and F. There was no nystagmus.

Vestibular and ocular motor responses

Smooth pursuit (SP, light dot stimulus at 20°/s), optokinetic nystagmus (OKN, black and white stripes of 7.5° width, stimulus field 120° in front of the patient, stimulus velocity

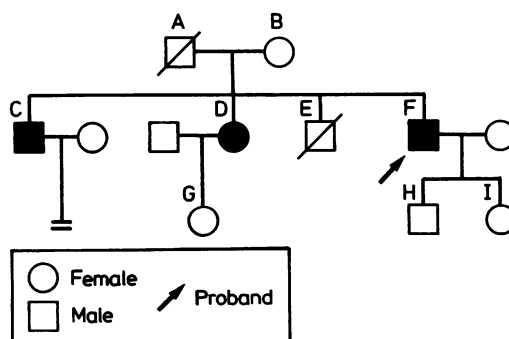


Fig 1 Pedigree of the family. (A) and (B) were dead. (C), (D) and (F) underwent the full examination; (G), (H) and (I) completed a checklist and a physical examination.

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20°, 40°, 60° and 80°/s) in both horizontal and vertical directions, and saccades (stimulus amplitude 20°) were undisturbed. Optokinetic after-nystagmus (OKAN) was absent in darkness (eyes open) after 60 s rotation at a constant speed of 40°/s in full light with eyes open.⁹ Using a Tonnie's rotatory chair, velocity steps of 90° or 180°/s were performed in either direction after acceleration at 0.8°/s in darkness.¹⁰ Neither per-rotatory nor postrotatory nystagmic responses were present. Sinusoidal vestibular stimulation in darkness (maximum velocity 50–100°/s, period 20s) did not produce any nystagmic response. Reading was impossible during sinusoidal horizontal head movements similar to the conditions used by Longridge and Malinson,¹¹ that is, about 1 Hz frequency and a maximum velocity of 100–120°/s.

Audiograms and brainstem auditory evoked responses (BAER; clicks with condensation- and rarefaction, 70 dB hearing level) were normal. Petros radiographs and brain CT scans in case (F) did not reveal any abnormality. Laboratory tests including autoimmune tests were normal.

Discussion

Blurred vision and illusory movements of the visual world during head movements are described by patients with oscillopsia often as very odd visual and vestibular experiences such as “bobbing”, “wobbling”, “dancing”, “jumping” and “jumbling” of the viewed object which “can't stand still”, “is all mixed up” or “is bouncing up and down”, shimmering and unsteadiness. On the other hand, pure vertigo, nausea and vomiting, well known and common symptoms in vestibular disorders, are mentioned less. Therefore one may not primarily think of defective labyrinthine function primarily, when seeing these patients. The vestibulo-ocular reflex (VOR) attempts to keep the image stable on the retina. Ideally passive or active head movements in one direction are compensated for by eye movements of equal magnitude in the opposite direction, so as to stabilise the gaze. In the case of vestibular stimulation, compensatory eye movements may be adequate up to 8 Hz.¹² Pursuit eye movements are adequate only up to a frequency of 1 Hz. Looking at a small fixation object normal humans do not report oscillopsia during 1 Hz sinusoidal rotatory head movements (amplitude +/–20°); oscillopsia is present in all normals at 2 Hz +/–20°.¹³ Wist *et al* used these findings in a simple clinical test; oscillopsia during head oscillation at +/–20° amplitude and a frequency below 1 Hz provides a reliable measure of pathological ocular motility.¹³ Disturbances in the VOR may result in oscillopsia. The differential diagnosis is important. The lesion may be found in the vestibular end organ or in the central nervous system, especially the brainstem and cerebellum (for review see ref 14). Our cases C, D and F did not show any abnormality other than absent VOR and OKAN; SP and OKN were normal. Absent OKAN is well known

in bilateral vestibular function loss.¹⁸ There was no hearing loss and BAEPs were normal. CT scans and petrosal radiographs were normal including the semicircular canals. Absence of lateral semicircular canals resulting in VOR dysfunction is one of the most frequent anomalies of the inner ear.¹⁵ There was no history of the use of any neuro-ototoxic drug, which may cause vestibular and cochlear function loss.^{2 14 16} Clinical manifestations or abnormal laboratory test results related to autoimmune disease were absent. Hughes *et al*¹⁷ tested the vestibular system in seven cases with autoimmune inner ear disease. They performed caloric and velocity step tests (100°/s). Five patients had ipsilateral or bilateral reduced or absent vestibular responses, one patient had positional nystagmus only and one case had normal responses. Two patients had a Dandy syndrome, that is oscillopsia and dysequilibrium when walking in the dark. All had sensorineural hearing loss on one or both sides. The same holds for the four patients presented by Belal² with (late onset) vestibular areflexia.

A congenital vestibular end organ dysfunction seems probable in the present patients. The pedigree suggests autosomal recessive inheritance. Blair Simmons⁵ described the findings in 43 patients with total bilateral absence of caloric responses. Five cases were considered probable or suspect unspecified congenital loss. One of these also had progressive hearing loss, another had a birth trauma; the three others had totally negative history and physical examination, as in our cases. No familial disorder was mentioned by these authors. Oscillopsia was the most common symptom, but this event was not always volunteered by the patients who had it. Two of our patients (C and D) had never asked for medical help and did not volunteer oscillopsia, probably owing to the fact that it was so common for them that they did not complain about it. Levin¹⁹ saw a 27 year old man with bilateral vestibular dysfunction without hearing loss. This patient suddenly noticed oscillopsia at 22 years of age which had continued unchanged. Laboratory tests were normal including cerebrospinal fluid examination. The aetiology of this case remained obscure, but it seems different from our patients. For revealing the cause of the disturbance in our cases it would be necessary to study suitable necropsy material. A developmental disorder of the inner ear vestibular sense organs (and/or vestibular nerves) however, seems probable.

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