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Clinical Pearls

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Climber exhibits first clinical manifestation of spinocerebellar ataxia on Karakoram expedition

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Case presentation

A 28-year-old man from Poland began to experience disturbances in gait and balance above 4150 m, while climbing one of the 7000 m peaks in Karakoram—the second highest mountain range on Earth (Figure 1). He struggled to coordinate his movements, which became noticeably slower. These symptoms increased gradually and were unaccompanied by disturbed consciousness, headache or vomiting.

He had never previously experienced these symptoms at sea level or at altitude, though he was an experienced climber. He was well-acclimatized to the 4150 m altitude (Figure 2) and reported no earlier symptoms that would have suggested altitude illness.

The climber had not taken medications for any condition before the expedition. His older brother had been diagnosed with autosomal dominant spinocerebellar ataxia type1 (SCA1) and the family was later found to have an undiagnosed multigenerational history of this condition. The mother, her sister and the grandmother presented similar symptoms. In his asymptomatic period the climber had been genetically tested and diagnosed with expanded CAG trinucleotide repeats in the ATXN1 gene.

One member of this Karakoram expedition was a doctor. In his opinion it was very likely that high mountain climbing triggered the manifestation of the pre-existing climber problem (hereditary ataxia). The affected man did not receive any pharmacotherapy at altitude. The symptoms prevented him from further climbing and he was forced to abandon the expedition. Descending from the base camp level, the climber suffered a fall and was finally transported down on a stretcher. The abnormalities persisted at sea level, though less severely.

On returning to Poland from Pakistan he was taken care of by a neurologist and had magnetic resonance imaging of his head, which revealed mild cerebellar atrophy and thinning of the brainstem—radiological findings in SCAs (Figure 3).

The final diagnosis of this cerebellar dysfunction was hereditary ataxia—SCA type 1. His symptoms have progressed ever since. The man underwent stem cell therapy in China, however it was not successful. After several years he needs help from others in his daily activities. The patient undergoes permanent rehabilitation.

Discussion

Ataxia is one of the well-documented disturbances at high altitude but some neurological conditions associated with this and other symptoms manifesting themselves at altitude might fall outside the usual definition of altitude sickness.[1,2]

Hereditary cerebellar ataxias are a genetically heterogeneous group of diseases with a similar clinical picture, whose course is most often slowly progressive.[3] Typical symptoms include disturbances in gait, coordination of movements, speech and in some forms, e.g. eye movement disorders.

Hereditary ataxias can reveal themselves from early childhood to late adulthood and might be inherited as autosomally dominant, recessive or X-linked, or mitochondrially.[4] They are rare diseases and so for example the global prevalence of SCA is 1–5 per 100 000; prevalence in Europe is 0.9–3 per 100 000 with significant geographical and ethnic variation.[5]

Inherited (genetic) forms of ataxia must be distinguished from the many acquired causes of this condition. Differential diag-

Figure 1. Photograph of Karakoram from the first author's archive.

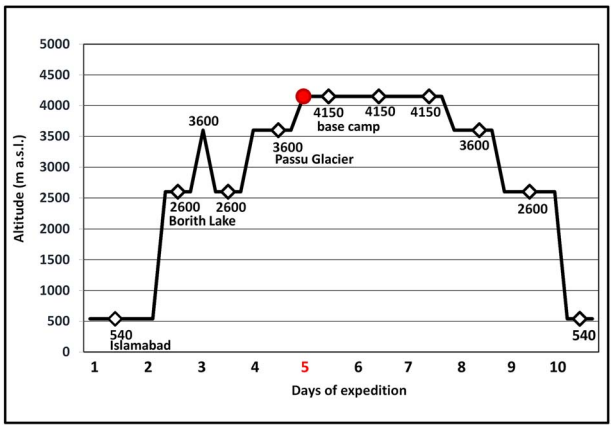

Figure 2. The rate of ascent of the described patient. Legend: white diamonds—sleeping altitude; red circle—the onset of symptoms.

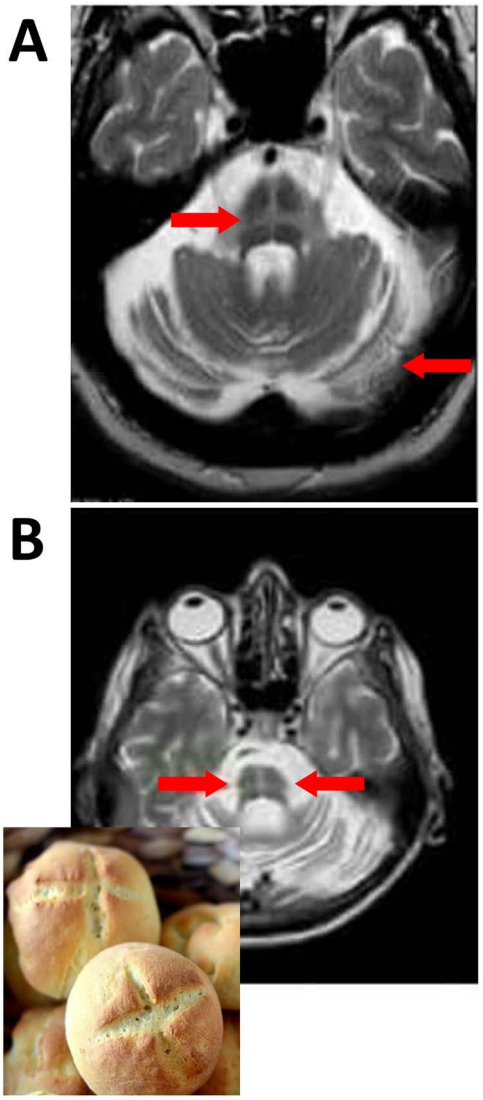

Figure 3. Magnetic resonance imaging of the patient's head showing several of the neuro-radiological findings (arrows) in the spinocerebellar ataxias, such as (A) cerebellar volume loss with (A + B) brainstem atrophy and the 'hot cross bun' sign.

nosis includes non-genetic causes of ataxia, such as alcoholism, vitamin deficiencies, multiple sclerosis, vascular diseases, primary or metastatic tumours, paraneoplastic syndrome associated with occult carcinoma of the ovary, breast or lung, toxic-induced and infectious cerebellar syndrome.[6] The genetic forms of ataxia are diagnosed by family history, physical examination, neuroimaging and molecular-genetic testing. The described patient fulfilled these conditions, and his family history of spinocerebellar ataxia and the result of a genetic test carried out in the asymptomatic period were very helpful in making an initial diagnosis at altitude.

Disturbances in gait, balance and coordination of movements experienced at altitude often suggest high-altitude cerebral edema (HACE). Ataxia, the impaired coordination of movements not resulting from paresis, is an important clinical finding in severe high altitude illness.[7] The most common symptoms of HACE, which is usually preceded by 24–48 hours of progressing acute mountain sickness, include: headache, severe lassitude, disturbed consciousness and ataxia.[8]

In this case, however, descending from height did not resolve the symptoms, which progressed over time, even at sea level.

The cause of ataxia occurring at altitude might be hypoxia affecting basal ganglia and hindbrain activity.[9] Remaining at high altitude might induce many disabilities, including neurological disorders. Exposure to high altitude with insufficient oxygen leads to many changes in the nervous system. Hypoxia causes a range of molecular, cellular and neuronal modifications and injuries. There is an increase in pressure in the brain's capillaries, disturbances in venous outflow, the release of various mediators, e.g. bradykinin, histamine, nitric oxide, increased activity of the sympathetic system or the action of vascular endothelial growth factor at altitude.[10] Due to all additional changes previous asymptomatic brain damage might begin to give symptoms.

Patients consent

The patient provided written consent for the publication of this report.

Conflict of interest

None declared.

Authors' contributions

Conceptualization was done by M.S.; Methodology was performed by R.K.S. and M.S.; Formal analysis was performed by R.K.S., M.S. and J.P.; Data curation was done by R.K.S. and M.S.; Original draft was written and prepared by R.K.S. and M.S.; Review and editing was done by R.K.S., M.S. and J.P.; Visualization was done by R.K.S. and M.S. ;Supervision was done by M.S.; Funding acquisition was done by R.K.S.

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