Orthostatic intolerance in two patients with Down syndrome

Abstract

Background: This article describes transient loss of consciousness and the diagnostic process in two patients with Down syndrome.

Methods/ results: One patient was diagnosed with initial orthostatic hypotension and postural tachycardia, the other patient was diagnosed with initial orthostatic hypotension.

Conclusions: Both patients were reassured about the cause and benign nature of their condition. People with Down syndrome could be at a higher than normal risk of episodes of TLOC and especially reflex syncope.

Key words: Down syndrome; transient loss of consciousness; reflex syncope; vasovagal syncope; orthostatic hypotension

Introduction

Transient loss of consciousness (TLOC) and orthostatic dizziness are common clinical problems. One third to half of the population experiences one or more episodes of loss of consciousness during their lives. Most of these episodes are caused by reflex syncope, of which vasovagal syncope, or fainting, is the most common form. Reflex syncope is caused by an inappropriate development of reflex arteriolar dilatation and cardiac slowing leading to arterial hypotension, reduced blood flow to the brain, and transient loss of consciousness. Other forms of reflex syncope are situational syncope, carotid sinus syncope and initial orthostatic hypotension.

People with Down syndrome have a low blood pressure compared to people without Down syndrome and frequently suffer from cardiac conditions and epilepsy. Therefore people with Down syndrome could have a higher than normal risk on TLOC and vasovagal episodes or (initial) orthostatic hypotension as a cause for their TLOC. At our outpatient syncope-department we recently saw two people with Down syndrome, one losing consciousness periodically and one complaining of light-headedness when rising.
Cases

Patient one is an eighteen-year-old male patient with Down syndrome. Since two years he suffers from episodes of TLOC about once a month, when standing, sitting or during activity. During these episodes he is faint and pale for about three minutes. During daytime he often falls asleep and snores. For his episodes of TLOC he was treated with Clonazepam, an anticonvulsive, which did not improve his complaints. The patient uses a wheelchair because of fatigue and pain in his joints. His medical history shows an Atrial Septal Defect, which was closed a year before presentation. Besides this he has recurrent airway infections with hearing loss and obstructive airway problems. He lives with his parents and two brothers.

The patient has a length of 1.60 metres and 52 kilos. On cardiac auscultation a normal first, and split second hearth sound, changing with breathing were heard. Otherwise physical and neurological examination were normal. The EEG showed no signs of epilepsy, sleep registration showed no sleep apnoea syndrome. On the ECG a sinusrythm of 160 bpm, and a right bundelbrachblock were seen. Twenty-four hour holter registration showed sinus rhythm with an AV-junctional rhythm of 50 bpm. Transthoracal echo-cardiogram showed a slightly dilated right ventricle with reasonable function, a dilated mitral valve, and slight mitral and tricuspid valve insufficiency. During cardiovascular reflex-examination, using a non-invasive finger arterial pressure monitor (Finapres Medical Systems) his supine blood pressure (BP) was 109/69 mmHg with a heart rate (HR) 60 beats per minute (bpm). Upon rising he showed a normal initial heart rate and blood pressure response. During 5 minutes of standing orthostatic adaption is normal, with an upper arm BP after three minutes of standing of 117/70 mmHg. When rising from squatting finger BP decreases from 130/102 mmHg to 85/50 mmHg, while the patient reports a strange feeling in the head. The history and induction of recognizable symptoms perfectly fit the diagnosis initial orthostatic hypotension, an explanation for his weight loss and reduced appetite was not found. He and his parents were reassured.

The second patient is a twenty-nine year old male with Down syndrome. He has complaints of light-headedness, especially when rising in the morning, and fatigue. He never really fainted. Recently he lost weight and has a reduced appetite. He uses Thyraz for his hypothyroidism. The patient lives at home, and used to work in a day care centre. During cardiovascular reflex examination his BP in supine position measured on the upper arm was 127/83 mmHg, with a finger BP of 105/60 mmHg, and HR of 85 bpm. Upon rising he showed a normal initial heart rate and blood pressure response. During 5 minutes of standing orthostatic adaption is normal, with an upper arm BP after three minutes of standing of 117/70 mmHg. When rising from squatting finger BP decreases from 130/102 mmHg to 85/50 mmHg, while the patient reports a strange feeling in the head. The history and induction of recognizable symptoms perfectly fit the diagnosis initial orthostatic hypotension, an explanation for his weight loss and reduced appetite was not found. He and his parents were reassured.

Discussion

Blood pressure is low in patients with Down syndrome. Average systolic blood pressure is 29 mmHg lower than that in general population. The cause of this lower blood pressure is not known sofar. Almost 10% of the people with Down syndrome are diagnosed with epilepsy. In case of TLOC due to low blood pressure significant convulsion-like limb jerking can occur, often erroneously interpreted as epileptic seizures. Syncope as a cause of TLOC can therefore easily be misdiagnosed as epilepsy, resulting in inadequate or inappropriate treatment as in our first patient. The prevalence and various causes of TLOC in people with Down syndrome is unknown. In subjects without Down syndrome, studies show that in about 25% of the patients diagnosed with epilepsy, this diagnosis is false or should be doubted seriously. In people with an intellectual disability this number could be even higher, because of the assumed high pre-test likelihood and descriptions of witnesses of events, and are commonly empirically treated with anti-convulsives. In people with an intellectual disability, history, cornerstone of diagnosing patients with TLOC, is often limited. Additionally, people with Down syndrome could also have a high risk on vasovagal episodes or (initial) orthostatic hypotension as a cause for their TLOC. They are known to have a significantly lower blood pressure than healthy controls, a high risk of (congenital) cardiac conditions, dementia, and can use cardiac or psychoactive medication. Knowledge about the prevalence of various causes of TLOC in people with Down syndrome and the value of history, physical examination, ECG and simple blood pressure tests therefore might raise the number of subjects with an adequate diagnosis for their TLOC-episodes, thereby receiving adequate treatment.
Conclusion

People with Down syndrome could be at a higher than normal risk of episodes of TLOC and especially reflex syncope. Orthostatic intolerance should therefore always be considered when patients with Down syndrome present with TLOC. To gain more insight in this problem studies on the prevalence of various causes of TLOC in this population should be performed.

Acknowledgments: none

Compliance with Ethical Standards:

Funding: none

Ethical approval: All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

References