Other full case

Acute altitude induced hypoxia in a child with Down's syndrome following postoperative repair of complete atrioventricular septal defect

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Summary

The authors report the case of a 4-year-old male child with a background of Down's syndrome (Trisomy 21) and a definitive repair of a balanced complete atrioventricular septal defect (CAVSD) at 3 months of age who experienced acute pulmonary oedema at high altitude (2000 m) while on holiday with his parents. The authors discuss and review the literature on the effect of altitude on children with Down's syndrome and postoperative CAVSD repair. The authors propose that further research is needed into this area in this specific group of patients, so advice can be given to families prior to flying or travelling.

BACKGROUND

Travel around the world, often to different altitudes, is now common. Children with chronic diseases often have to adapt and respond differently to various physiological conditions such as temperature, exercise and altitude for instance. A specific example we report is in the case of Down's syndrome. It is well documented that children with Down's syndrome have abnormal pulmonary vascular resistance and raised pulmonary pressures.¹ When this physiology is considered in a high-altitude setting where P_{aO_2} is lower, there may be an increase in the risk of high-altitude pulmonary oedema. We therefore report this case to raise awareness of the pulmonary vascular differences in children with Down's syndrome and to encourage people to consider the effect of altitude on the physiology of the patient they are dealing with.

CASE PRESENTATION

A 4-year-old boy with Down's syndrome (Trisomy 21) was diagnosed at birth as having a balanced complete atrioventricular septal defect (CAVSD) as part of the routine echocardiogram performed as per the Down Syndrome Recommendation group guidelines.² He underwent uneventful CAVSD corrective repair at the age of 3 months with only transient pulmonary hypertension postoperatively. Postoperative saturations ranged from 95% to 100% in room air. At no point prior to his repair did he have symptoms of heart failure or require any treatment related to heart failure. He remained well with no significant medical history until a problem that arises while on a holiday.

He and his family flew to Geneva, followed by a coach trip to Belle Plagne, France. The coach ascended to an altitude of just over 2000 m over a 3-h period. Parents report that the following day they noticed that he appeared tired and had lost his appetite, but initially felt this was due to the long travel and very late arrival into Belle Plagne. On day 2, at this altitude he remained drowsy with intermittent

periods of agitation. His oral intake decreased rapidly and his respiratory effort increased. His parents became concerned regarding a bluish discolouration of his lips. He was seen by a local doctor on the mountain range who documented his respiratory rate to be 60 breaths/min, his heart rate 190 beats/min and his saturations to be 66–68% in room air. The doctor checked the saturation probe on his own finger, and found his saturations to be 94% at this altitude. A concern was raised at this point that his drowsy and agitated state maybe secondary to cerebral oedema due to the altitude, but he was equally hypoxic and this in itself could lead to this presentation of behaviour.

The decision was made to transfer him down to sea-level again to the nearest hospital in Albertville. As the descent was made, he was noticeably more alert and in A&E at the hospital, his saturations increased to 88% in air at sea-level. A chest x-ray performed the following day was reported by the radiologists as showing pulmonary oedema. He was monitored for 48 h in hospital, with the initial 24 h on oxygen and he made a rapid improvement and was discharged to a lower altitude (1500 m). No diuretics were given due to his rapid improvement, simply by descending to sea-level. He was advised to return to an altitude no higher than 1500 m for the rest of his stay which was uneventful. No echocardiogram was performed when he was unwell at high altitude, but prior to his holiday and since return his pulmonary pressures have always been reported to be normal.

TREATMENT

He responded well on returning to sea-level and oxygen therapy. He did not require any other pharmacological treatment.

OUTCOME AND FOLLOW-UP

Since this episode, he has suffered an episode of pneumonia while in France (this time at altitude of 1500 m) and his

saturations rapidly decreased to mid-60%. His parents are now reluctant to ascend any higher than this. He has had several echocardiograms performed by the paediatric cardiology service at sea-level and all have been normal, with good function, no atrioventricular valve regurgitations and normal pulmonary pressures.

DISCUSSION

This case demonstrates that there is a high chance that the pulmonary vascular resistance in him is highly variable, given the rapid change in his saturations based on altitude and intercurrent illness. Unfortunately, echocardiogram data during the periods of hypoxia was not obtained.

Cardiac catheterisation data suggests that children with Down's syndrome and CAVSD have an abnormally high pulmonary arterial pressure when compared to CAVSD in children with a normal karyotype.¹ Furthermore, children with Down's syndrome have been demonstrated previously to have increased risk of high-altitude pulmonary oedema. Many reasons have been speculated such as underlying chronic pulmonary hypertension (sometimes secondary to obstructive sleep apnoea), previous pulmonary damage from high left to right cardiac shunts or frequent infections. Repair of the cardiac lesion does not seem to alter the children's susceptibility to this phenomena.³ In view of the altered vascular reactivity along with multiple reasons for raised pulmonary arterial systolic pressures, it is not surprising that children with Down's syndrome are at risk from the affects of altitude.

The pathogenesis of high-altitude pulmonary oedema in all age groups has had much speculation. In fit and well individuals small accumulation of fluid in the alveoli has been demonstrated to be a normal effect at high altitudes. In normal circumstances this should be cleared quickly. However, this depends on the ability for the pulmonary vascular bed to be able to alter its resistance in a suitable manner, often using substances such as nitric oxide in this process.⁴ Furthermore, children with Down's syndrome have been demonstrated to have a reduced total number of alveoli, and the alveoli that are present are often larger with a degree of pulmonary hypoplasia.^{5 6} If a child with Down's syndrome is exposed to this alveolar fluid and then becomes hypoxic, thus worsening the pulmonary vascular

resistance, and taking into account all the above factors, it can be seen how they can become acutely unwell with pulmonary oedema.

From this case, we propose that parents of children with Down's syndrome with or without an associated congenital heart defect are given advice about the affect of altitude with regards to holidays. We suggest further research needs to be undertaken to review the affect of altitude in children with Down's syndrome and those with congenital heart disease in more detail.

Learning points

- ▶ Children with Down's syndrome and CAVSD may have abnormally high pulmonary arterial pressures, for a variety of reasons.
- ▶ Children with Down's syndrome have increased risk of acute high-altitude pulmonary oedema and this should be considered as a diagnosis in areas of high altitude.
- ▶ Consider giving advice to parents regarding slow ascent (if possible) and clinical features to observe for in their child when going on holiday, if at risk of high-altitude pulmonary oedema.
- ▶ Returning to sea-level can make a significant difference in the management of high-altitude pulmonary oedema.

Competing interests None.

Patient consent Obtained.

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