Cerebrospinal Fluid Research



Oral presentation

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Hypoventilation and sleep apnoea in spina bifida - an unrecognised problem?

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from 53rd Annual Meeting of the Society for Research into Hydrocephalus and Spina Bifida Belfast, UK. 24-27 June 2009

Published: 27 November 2009

Cerebrospinal Fluid Research 2009, 6(Suppl 2):S39 doi:10.1186/1743-8454-6-S2-S39

This abstract is available from: http://www.cerebrospinalfluidresearch.com/content/6/S2/S39

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Background

Hypoventilation and sleep apnoea syndromes are associated with increased body mass index (BMI) and Chiari malformation, both of which are common in the spina bifida (SB) population. Despite a number of published case reports, recent literature on the management of SB does not specifically mention such conditions. We discuss the presenting symptoms, aetiology, investigation and outcomes of our affected patients.

Materials and methods

Chart review of patients considered for ventilatory support (n = 6), identified from the database.

Results

Patients complained of a combination of problems, including respiratory symptoms, resistant lower limb oedema, and persistent erythrocytosis. Episodes of unresponsiveness and headache were also noted, and hydrocephalus/shunt malfunction was considered a differential in some cases. Five patients had shunts in place, and four patients had undergone scans confirming Chiari malformation.

Following appropriate investigation, including pulmonary function testing and sleep studies, three patients were commenced on nocturnal ventilatory support, with symptomatic improvement. The remainder are under regular review by the respiratory physicians with a view to future treatment.

Conclusion

A low threshold for further investigation and referral should be employed in SB patients with symptoms suggestive of respiratory dysfunction. Those felt appropriate for nocturnal ventilatory support can have significantly improved Quality of Life and symptom resolution with treatment. Both investigaton and treatment are non-invasive and widely available.

This is particularly important where symptoms could be attributed to Chiari malformation or decompensation of arrested hydrocephalus, as the patient may avoid the risks attendant to neurosurgical procedures.