Chiari type I malformation in two adult patients with cystic fibrosis

D. Narasimhaiah1, C. Etherington1, G. Fitch1, P. Whittaker1, D. Peckham1. 1St James’s University Hospital, Regional Adult Cystic Fibrosis Unit, Leeds, United Kingdom

Objectives: The Cystic Fibrosis (CF) gene is associated with abnormal production and function of the cystic fibrosis transmembrane conductance regulator (CFTR). Despite the widespread expression of CFTR in the brain, neurological complications and malformations of the central nervous system (CNS) are rare. We report two cases of Chiari malformations in adults with CF.

Methods: Case records were reviewed. Diagnoses were confirmed by brain MRI scan and neurological assessment.

Results: Since 2006, 440 patients (≥17 years) have been followed up in the adult unit. Two patients were diagnosed as having Chiari Type I malformation.

The first case involved a 26 years old female who presented with secondary amenorrhea due to hyponagadotrophic hypogonadism. Brain MRI demonstrated Chiari type I malformation with hydrocephalus requiring a third ventriculostomy and decompression of foramen magnum. The second patient was a 23 year old male who presented with type II respiratory failure and persistent headache which failed to respond to non invasive ventilation. Brain MRI confirmed Chiari I malformation without evidence of hydrocephalus.

Conclusion: Chiari type I malformation is characterized by herniation of the cerebellar tonsils through the foramen magnum and can be either asymptomatic or present with a wide spectrum of neurological symptoms. Although rare (1/1000 births), Chiari type I malformation may be more frequent in patients with CF [1].

The diagnosis should be considering in patients presenting with neurological symptoms.

Reference(s)

Aspergillus nodi in cystic fibrosis (CF) patients: a rare entity. Report of two pediatric cases

P. Schelstraete1, S. Van Daele1, F. Haeryna1, F. De Baets1. 1Ghent University Hospital, Cystic Fibrosis Reference Center, Ghent, Belgium

Introduction: Aspergillus nodi are rarely reported in CF, mostly in adult patients with CF-related diabetes and severe pulmonary damage (FEV1 <25%) and/or under immunosuppressive treatment. This complication is rarely seen in children. We report two CF children with aspergillus nodi.

Cases: Patient 1 presented at the age of 13 y with dyspnoe, thoracal pain and cough without fever. Chest X-ray and HRCT revealed a round pneumonia in the right upper lobe. A. fumigatus was cultured of bronchoalveolar lavage (BAL) fluid, BAL galactomannan was positive. A complete cure was obtained after a 6 month treatment of orally administrated antifungals (voriconazolet, switched to itraconazol because of side effects).

Patient 2 was seen on the outpatient department at the age of 9 y because of pain during inspiration and a mild cough without fever. Lung HRCT scan revealed a nodular lesion in the right upper lobe. BAL fluid culture was positive for A. fumigatus, BAL galactomannan was positive. Treatment consisted of oral voriconazol during 6 months, after which complete healtment was obtained.

Conclusion: Although invasive infections by A. fumigatus are preferentially but rarely seen in CF adults with underlying conditions, aspergillus nodi should also be considered as a rare complication in CF children without predisposing factors.

Pseudomonas aeruginosa colonization in a CF child’s upper and lower airways: a father’s meticulous approach to successful eradication

A. Gerber1, T. Ebener1, M. Lorenz2, J. Hentschel1, W. Pfister2, J.F. Beck1, J.G. Mainz1. 1Jena University Hospital, CF-Centre, Jena, Germany; 2Jena University Hospital, Institute of Medical Microbiology, Jena, Germany

First Pseudomonas aeruginosa (PSA) colonization of the presently 8-year-old male patient with CF was diagnosed by a routine deep throat swab. Subsequently both, the upper and lower airways (UAW/LAW) were sampled monthly by throat swabs/sputum collection, and nasal lavage, according to the Jena CF-centers’ standard for intermittent colonization. The boy’s father meticulously documented all therapies and culture results.

Initial attempts to eradicate PSA following current German guidelines, with oral ciprofloxacin, inhaled colistin and tobramycin, as well as three cycles of IV cefazidime and tobramycin failed. After the first positive nasal sample, vibrating nasal inhalation of nebulized colistin (Pari Sinus) was added to the regimen. After 9 positive cultures of PSA from the LAW and 3 from the UAW, we changed to an off-label regimen consisting of inhaled aztreonam alternating monthly with tobramycin (given simultaneously for UAW and LAW, and IV antibiotics every 3 months, and oral azithromycin).

Our patient has now been free of PSA for one year, as documented by monthly negative cultures. This case demonstrates that routine sampling of lower and upper airways may reveal early nasal colonization, which if unrecognized, would have precluded successful LAW eradication. Monthly sampling detected intermittent colonization and confirmed complete clearance of PSA. Both would not have been achieved following international standards for CF care, which only recommend LAW sampling, 2–4 times/year. Bronchial antibiotic inhalation alone would likely have missed treating the UAW. The whole process was closely monitored by the boy’s father. We want to thank him for his commitment.

Comparison of airway clearance techniques (ACT) in the cystic fibrosis (CF) population: a single case study report

E. Forster1, E. Swingwood2, B. Nicholas1, K. Bateman1. 1UHBristol NHS Foundation Trust, Bristol Adult Cystic Fibrosis Centre, Bristol, United Kingdom

Objective: Airway clearance techniques (ACT) are an integral component of CF care, although the most effective method is debated. Non-invasive ventilation (NIV) and Intermittent Positive Pressure Breathing (IPPB) are commonly used. Mechanical Insufflation:Exsufflation (MI:E) may provide a useful alternative. To compare ACT performance we reviewed flow and pressure breath profiles and regional lung ventilation (RLV) via Electrical Impedance Tomography (EIT).

Method: IPPB (Pressure 28–30 cmH2O), NIV (28/6) and MI:E (Insufflation +28 cmH2O:Exsufflation −28 cmH2O) were used in 3 separate ACT sessions over a 2 week period in 1 CF patient. Constant monitoring of breath profiles and RLV via EIT occurred. A 10-point visual analogue scale (VAS) for ease of sputum clearance was recorded pre and post treatment.

Results: RLV increased during all ACT from baseline and was significant for MI:E and NIV (p <0.05). NIV breath profiles suggested limitation of expiratory flow in comparison to the exsufflation breath of MI:E and IPPB breaths, where an expiratory flow bias was apparent. Expiratory flow bias is essential for effective ACT, preventing embedding of mucus. MI:E was reported as most effective ACT by the patient, with VAS for ease of clearance reducing from 6 to 3.

Conclusion: Lung recruitment occurred with all ACT. NIV may limit expiratory flow and sputum clearance. IPPB and MI:E may facilitate sputum clearance due to expiratory flow bias. Patient-reported VAS for ease of sputum clearance favored MI:E. This is a single case study report; further research may be of benefit to further investigate the potential use of MI:E in CF.