

CEREBRAL ABSCESS IN ADULT CONGENITAL HEART DISEASE (ACHD): A SINGLE CENTRE EXPERIENCE

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Background Cerebral abscess is a rare but recognised complication in cyanotic congenital heart disease (CHD) with a poor prognosis, especially if diagnosis is delayed¹. Method: Retrospective analysis of paper and electronic records of ACHD patients at Leeds General Infirmary (LGI) with a history of confirmed cerebral abscess.

Results 7 cases of cerebral abscess were managed in LGI 1981–2019 (table 1). One occurred prior to transition to adult services, mean age 34 years. All had cyanotic CHD, and all but 1 occurred prior to any cardiac intervention. All presented with either focal neurology or headache, with other symptoms in graph 1. Only 1 had normal inflammatory markers on admission and there was no evidence of vegetations on any echocardiograms. One patient had suture diastasis on skull x-ray, and all had a CT scan confirming ring enhancing lesions. One had multiple lesions and the location of the lesions was varied (parietal, frontal, thalamic and occipital). Three (43%) had streptococcus milleri on abscess aspirate with another documented as gram positive cocci. One had strep intermedius on blood cultures (member of streptococcus milleri group), but all other blood cultures demonstrated no growth. All underwent aspiration or surgical drainage of the abscess, 3 occurred on the day of diagnosis, and the longest duration prior to intervention was 13 days. All received a prolonged course of antibiotics of varying combinations (graph 2). Two developed seizures post-operatively and one developed a renal abscess. One patient had long-term neurological sequelae (Epilepsy and left homonymous hemianopia), one died 8 days post-operatively and another died 3 years later from end stage heart failure. Discussion: Streptococcus milleri group are recognised commensals of the oropharynx and gastrointestinal tract, and the predominant microbiological diagnosis in cerebral abscesses¹. Yet only one case had recent dental work, and none had confirmed endocarditis by Duke's criteria. Despite documented high rates of peri-operative complications², in our cohort there was one death and one long-term neurological sequelae. It is imperative the diagnosis is confirmed at the earliest opportunity, with prompt neurosurgical referral regardless of underlying cardiac pathology and degree of cyanosis as these high risk patients have demonstrated good long-term post-operative outcomes. We acknowledge the limitations that paediatric cases were not reviewed, and that clinical practice has progressed since the earliest case in 1981.

Conclusion Prognosis has improved in recent decades, but CHD remains a recognised risk factor for death³. It is an important differential to exclude in any patient with cyanotic CHD presenting with a headache or focal neurology by performing urgent cerebral imaging⁴. Clinical management relies on retrospective studies and previous clinical experience as no trials report clinically meaningful outcomes for antibiotic use in cyanotic congenital heart disease with cerebral abscess owing to the rarity of the condition⁵.

Conflict of Interest None

UTILISATION OF GENETIC TESTING IN AN ADULT CONGENITAL HEART DISEASE CLINIC

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Introduction A significant proportion of adults with congenital heart disease (CHD) have an underlying genetic diagnosis which can have important medical, reproductive, and psychosocial implications for these patients. In view of advances in genetic diagnostics, current best practice guidelines (1) recommend genetic testing in adults with CHD and additional clinical features which increase the likelihood of an underlying genetic diagnosis. We sought to ascertain the utilisation of genetic testing in an adult congenital heart disease clinic.

Methods We retrospectively reviewed the electronic clinic records of 102 consecutive patients attending the adult congenital heart disease clinic between March and June 2021. Data collection included the underlying congenital diagnosis and the presence or absence of anatomical or phenotypic features associated with increased genetic risk, in line with international recommendations for genetic testing in adult CHD: presence of a conotruncal abnormality (to include tetralogy of Fallot, ventricular septal defect with aortic arch anomaly, truncus arteriosus and interrupted aortic arch), recognisable extra-cardiac syndromic features, learning disability, developmental delay, psychiatric diagnosis, and family history of congenital heart disease.

Results 102 electronic patient records were reviewed. The mean age was 37.5 years (SD = 14.0). The most common congenital diagnoses were atrial septal defect (n=15), tetralogy of Fallot (n=16), and coarctation of the aorta (n=10). 11 patients had a confirmed genetic diagnosis. 2 patients were identified as having phenotypic features of a genetic syndrome with referral for testing in place. 7 patients had developmental delay or learning disability without an established genetic diagnosis. In total 23/102 (22%) patients, who had not been tested, met criteria for genetic testing.

Conclusion Genetic testing was underutilised in this cohort of patients. It is likely that a significant number of patients with an underlying genetic diagnosis are being missed, particularly as some syndromic diagnoses may have subtle features which may not be apparent on evaluation in clinic. Patients who would benefit from genetic testing need to be pro-actively identified in adult CHD clinics to ensure patients are optimally clinically managed and receive appropriate genetic and pre-conception counselling.

REFERENCES

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