CONGENITAL BRAIN MALFORMATIONS IN NEONATES WITH CONGENITAL HEART DISEASE

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INTRODUCTION

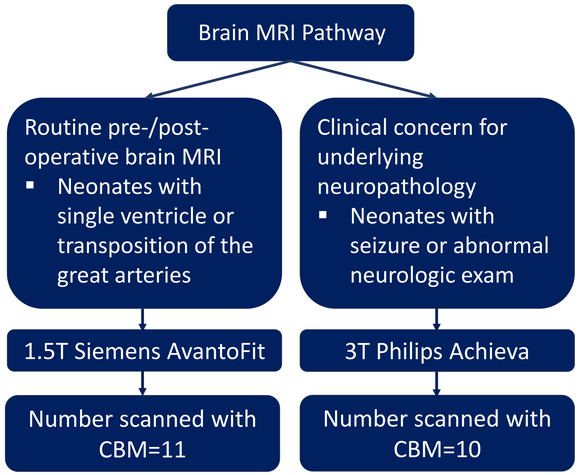
- Neonates with congenital brain malformations (CBM) have historically been excluded from neuroimaging studies in the congenital heart disease (CHD) population

OBJECTIVES

- The objective of this study is to describe CBMs, associated genetic diagnoses, and impact on clinical care in neonates with CHD

METHODS

- This single-centre retrospective study included neonates with CHD born between January 2018-June 2023 diagnosed with CBM on MRI



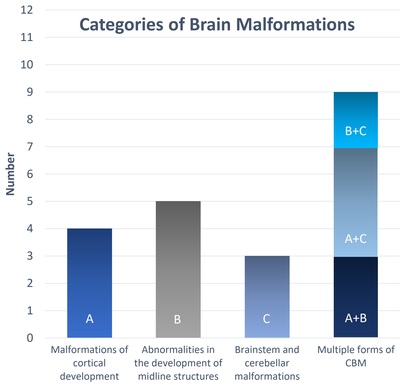
RESULTS

1 *Clinical characteristics vary across the population*

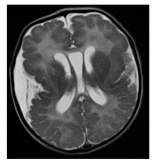
Characteristics	n=21
Male	12 (57%)
Birth gest. age (weeks)	38.1 (30.2-40)
Birthweight (g)	2780 (1380-4110)
Head circumference at birth (cm)	33 (27.8-37)
Cardiac Diagnosis Categorization	
Single ventricle physiology	6 (28%)
Biventricular physiology w/o arch obstruction	11 (52%)
Biventricular physiology w/ arch obstruction	5 (24%)
Prenatal cardiac diagnosis	16 (76%)
Prenatal CBM diagnosis	5 (24%)
Maternal age	30 (21-47)
Maternal exposures during pregnancy	5 (23.8%)
Maternal infection during pregnancy	0 (0%)
Presence of dysmorphic features at birth	8 (38%)
Other organ system involvement (excluding heart/brain)	8 (38%)
Amniocentesis	5 (24%)
Post-natal genetic testing	20 (95%)
FISH	4/20 (20%)
Chromosomal microarray	18/20 (86%)
Whole genome/exome sequencing	11/20 (55%)
Seizures	6 (28.5%)
Deceased	5 (23.8%)

2 *Brain malformations in the cohort presented in three main categories*

CBMs were identified in 21 of 438 (4.8%, 95% CI 3.1-7.3) neonates who received cardiac care during this period

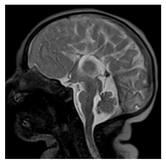


Malformations of cortical development



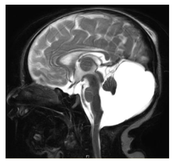
Axial T2 weighted sequence demonstrating severe polymicrogyria in a term neonate with single ventricle physiology

Abnormalities in the development of midline structures



Sagittal T2 weighted sequence demonstrating agenesis of the corpus callosum and simplified gyral pattern in a term neonate with biventricular physiology without arch obstruction

Brainstem and cerebellar malformations



Sagittal T2 weighted sequence demonstrating cerebellar hypoplasia and large retrocerebellar cyst in a term neonate with single ventricle physiology

3 *Diagnosis of a congenital brain malformation impacted clinical investigations and care*

75%
A genetic diagnosis was found in 15/20 neonates (75%, 95%CI 52.8-89.2)

30%
Other findings – 6/20 had a chromosomal anomaly

45%
Other findings – 9/20 had a pathogenic variant in a single gene

55%
Diagnosis of CBM on MRI prompted genetic investigations in 11/20 neonates

29%
6/21 neonates underwent palliation following diagnosis of the CBM

CONCLUSIONS

- CBMs in neonates with CHD are rare
- Early diagnosis of CBMs with MRI was important for the initiation of genetic investigations and for directing goals of care