

Obstetric and neonatal outcomes of pregnancies diagnosed with Foetal Cystic Hygroma anomaly in Bradford

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Background

- A foetal cystic hygroma (FCH) is a congenital malformation that presents as fluid filled sacs in the lymphatic system (**Figure 1**)¹
- The prognosis of cystic hygromas depends on the stage of diagnosis, with those identified in the first trimester displaying poorer prognoses²
- Literature suggests around 50% of cases are associated with a chromosomal abnormality³ and most fetuses experience a poor outcome
- The neonatal and paediatric outcomes of these babies are less widely followed up which restricts patient counselling.
- Diverse outcomes of FCH create uncertainty around the prognosis of this diagnosis which is challenging for physicians to communicate

Objectives

- To understand the prevalence of FCH in patients attending Bradford Royal Infirmary, England
- To conclude the main chromosomal and structural anomalies associated with a FCH diagnosis
- To identify the main obstetric and neonatal outcomes of a pregnancy affected by FCH

The overall aim was to gather new information suggesting prognostic indicators to guide prenatal and antenatal patient counselling on a FCH diagnosis.

Methods



Figure 1- an ultrasound scan displaying foetal cystic hygroma⁴

This research was a retrospective study conducted at Bradford Royal Infirmary (BRI)

Foetuses presenting with FCH were identified from consultant ultrasound MDT reports between 2017-2021

Electronic patient records were used to collect data on the mother, foetus, and pregnancy

Surviving babies were followed to conclude neonatal and paediatric outcomes

Results

FCH in Bradford

- 46 foetuses from 44 pregnancies were identified with FCH at BRI
- Prevalence averaged at 1 case per 588 pregnancies
- 31.8% of FCH cases were babies conceived through consanguineous relationships- all between 1st and 2nd cousins
- 2/3 of women had experienced a previous pregnancy loss

Chromosomal anomalies

- 16/44 (36.4%) of mothers declining invasive pre-natal testing
- There were 4 findings from chromosomal analysis (**Figure 2**)
 - 5/28 (17.9%) of pregnancies had a normal karyotype
 - Trisomy 13 was found in 4/16 (25.0%)
 - Trisomy 18 in 4/16 (25.0%)
 - Trisomy 21 in 6/16 (37.5%) and turners' syndrome in 2/16 (12.5%) of cases

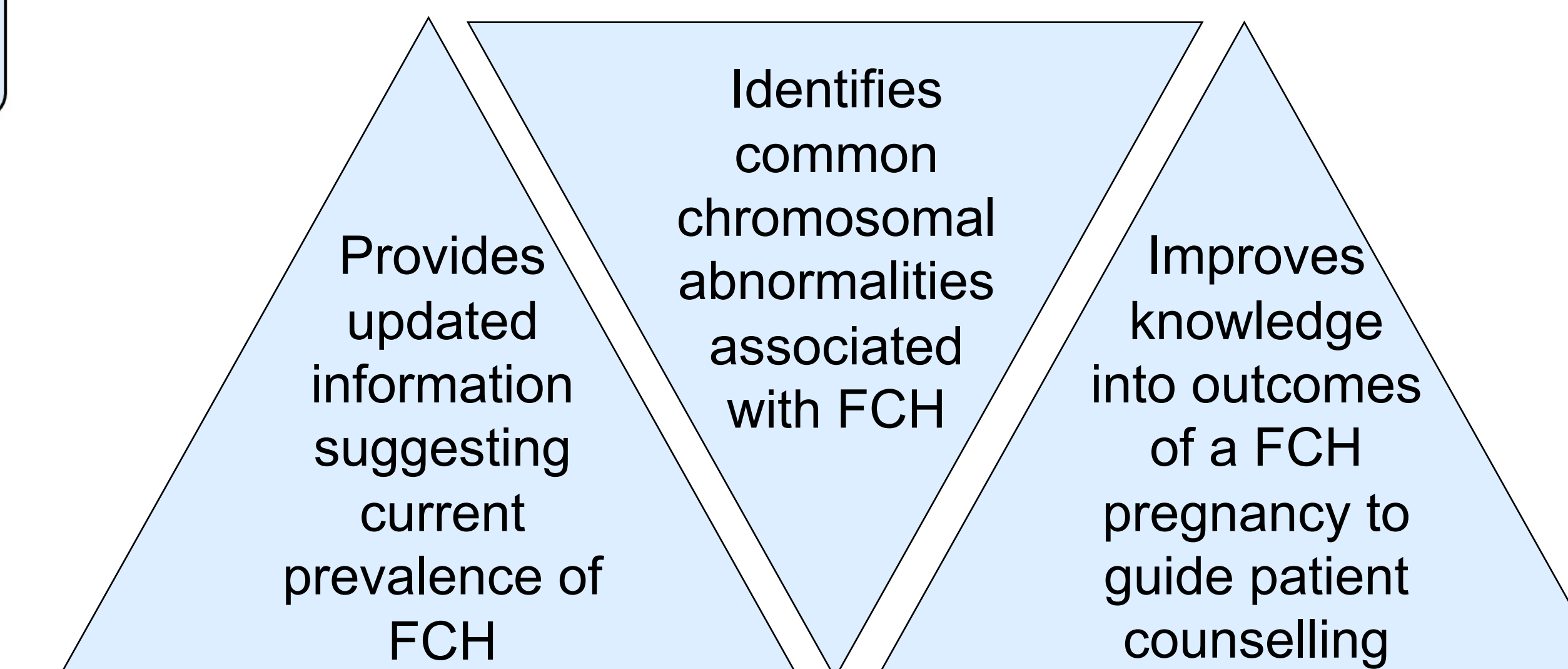
Obstetric outcomes

- 4 significant obstetric outcomes of a pregnancy affected with FCH were concluded, as displayed in **Figure 3**: previable loss, stillbirth, termination and live birth
- Of the IUD's that had consent for genetic testing, 100% (5/5) of conclusive results were positive for aneuploidies.
- All 3 cases of trisomy 21 were miscarried before 24 weeks.
- Slightly more foetuses died in the first 2 trimesters of pregnancy- only 3/11 (27.3%) losses occur in the 3rd trimester.

Conclusions

- Prevalence of FCH is around 1/588 pregnancies
- There is a strong association of FCH with consanguinity between parents
- FCH is strongly linked to chromosomal abnormalities including trisomy 13, trisomy 18, trisomy 21 and tuners syndrome
- Subsequent diagnosis of trisomy 21 is associated with higher rates of intra-uterine death
- Foetuses with normal karyotype are more likely to have a live birth and normal neonatal outcomes

Research benefits



References

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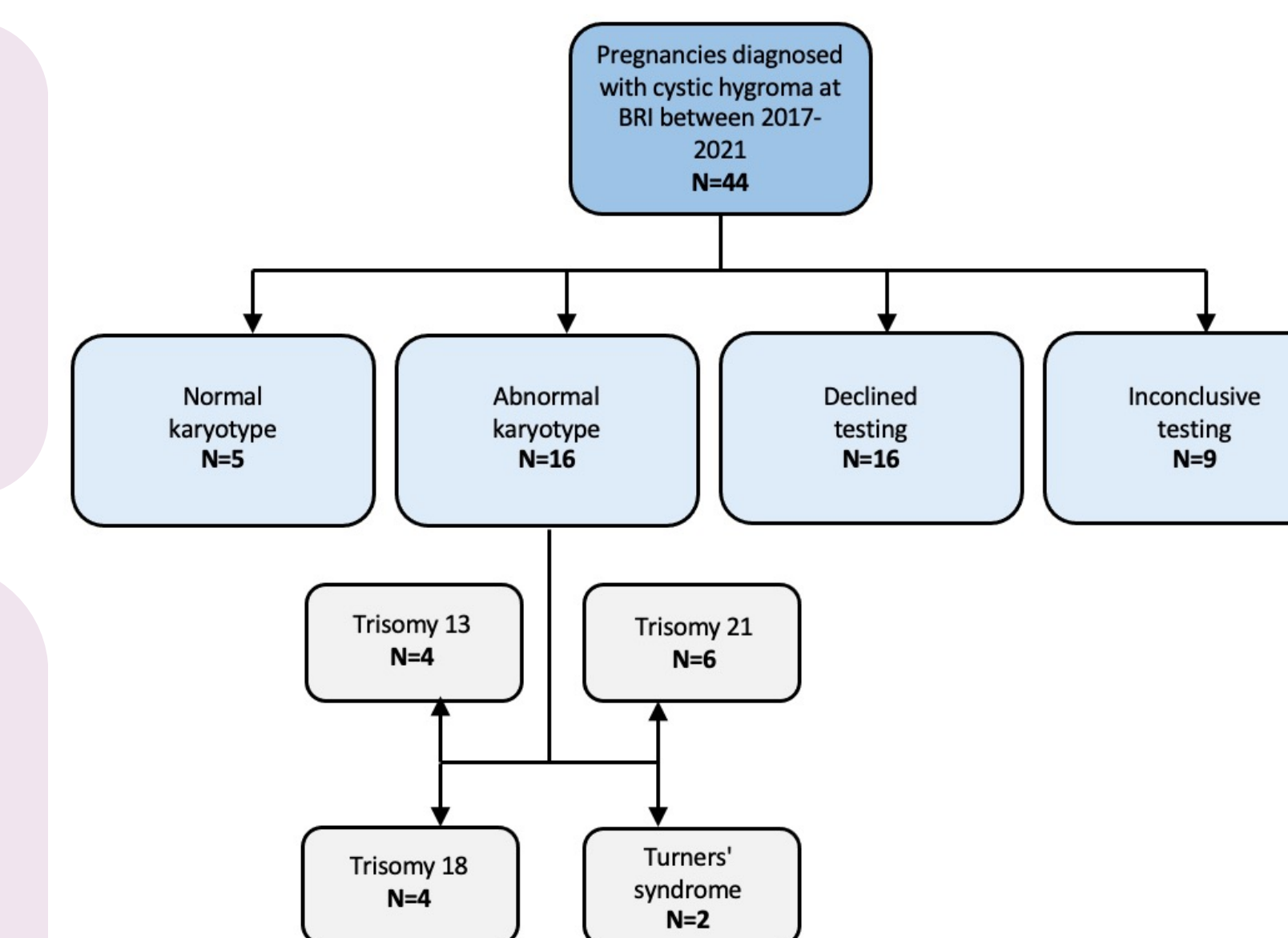


Figure 2- a diagram displaying karyotypes of foetuses diagnosed with cystic hygroma at BRI

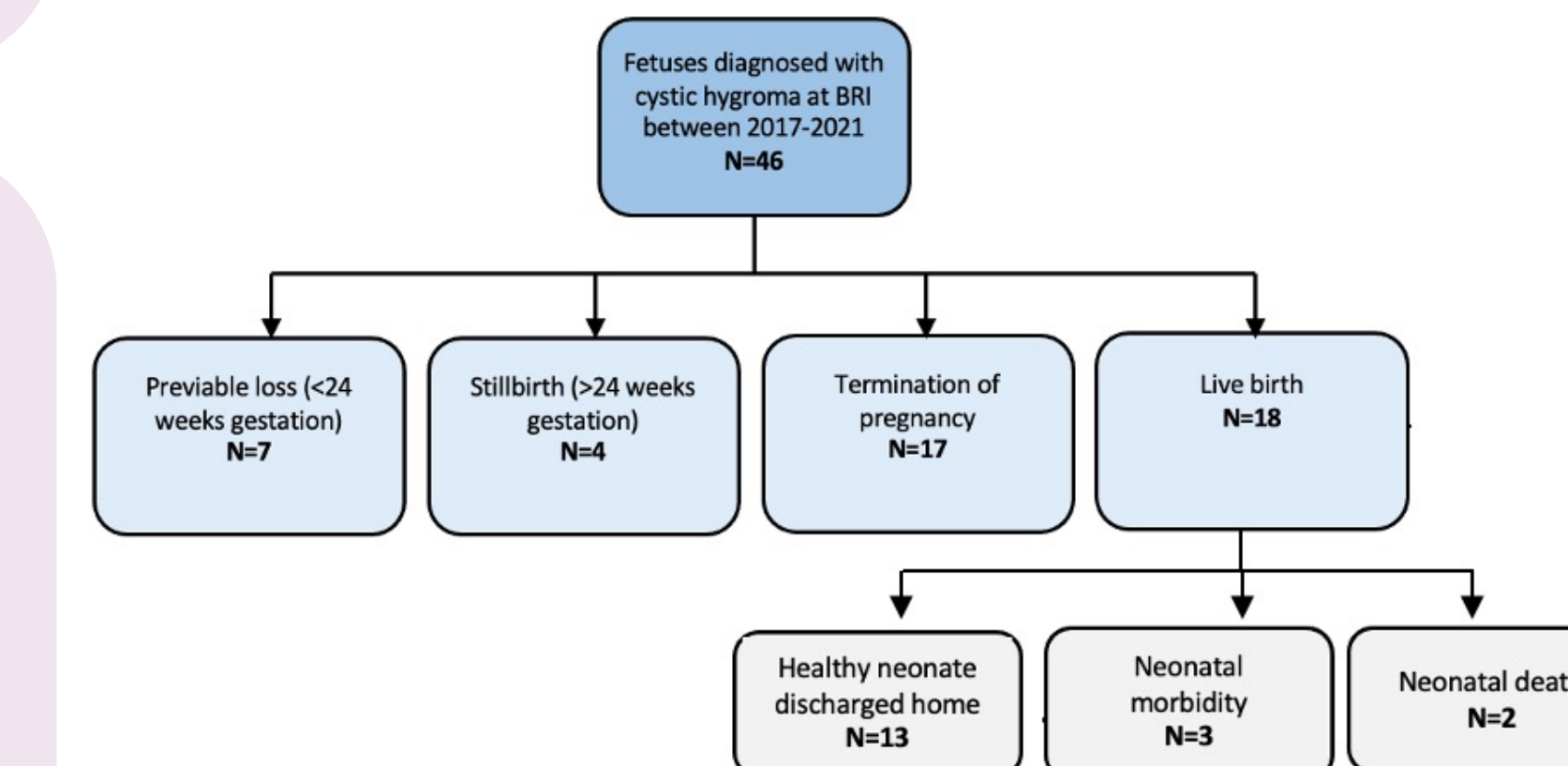


Figure 3- a diagram displaying obstetric outcomes of foetal cystic hygroma pregnancies at BRI