UNIVERSITY OF CAMBRIDGE

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The Community Child Health care of children with genetic diagnoses

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BACKGROUND

Many children under the care of Community Child Health Services (CCS) have genetic conditions, but for most genetic conditions there are no established care pathways.

The Next Generation Children's Project (NGCP) demonstrated clinical utility of trio whole-genome sequencing (WGS) in our regional NICU and PICU[1]. For children who received a genetic diagnosis through the NGCP, we examined CCS input with the following aims:

1. What proportion of children meet the threshold for CCS referral at diagnosis or likely to within the next year
2. Whether these children were referred to and seen by CCS
3. Whether appropriate educational support is in place for those children who seen by CCS

METHODS: A retrospective clinical audit

521 families received trio WGS through NGCP

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36 children received a genetic diagnosis and lived within catchment area

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1 patient excluded: incidental diagnosis not anticipated to manifest until later life.

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Data collection using hospital and community records

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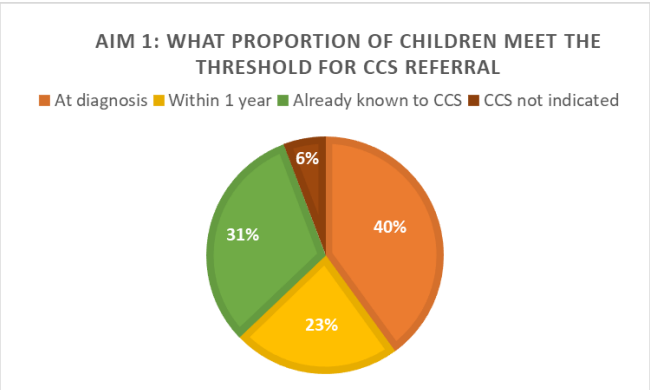
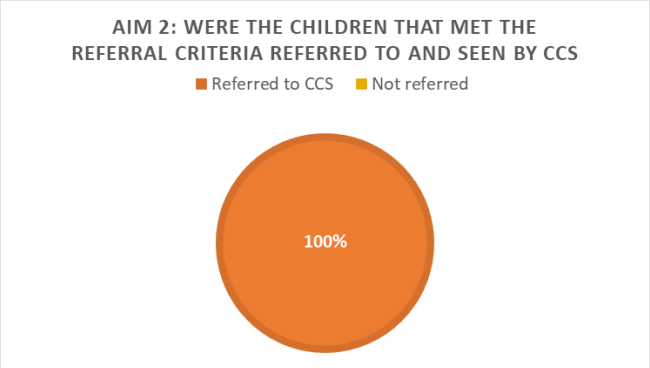
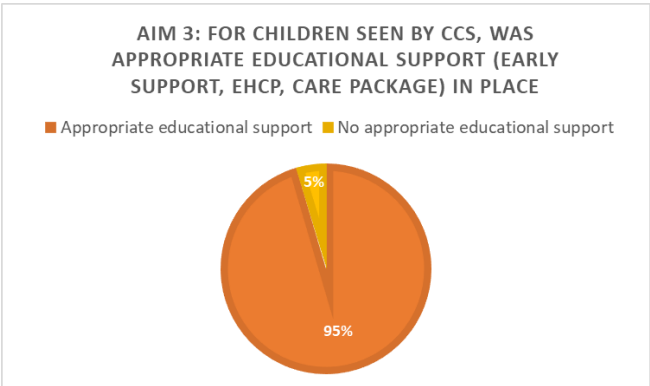
8 children excluded from analysis as referral to CCS not applicable
(6 died prior to discharge and 2 left the UK)

RESULTS





Patients known to CCS were significantly older ($p<0.05$, two-tailed T-test) at genetic diagnosis (mean age 4 years), than new referrals to CCS (mean age 4.6 months at genetic diagnosis). New referrals were received several months (mean = 7 months) after diagnosis



CONCLUSION

- The majority of children with a monogenic condition (63%) required input from CCS
- Children not known to CCS at the time of diagnosis tended to be infants
- Further work is required to explore whether parents would welcome an initial contact at the time of diagnosis to outline the support available and advice on how to access the relevant services as and when they are needed
- This is currently offered in our area for other genetic conditions such as Down's Syndrome

1. French CE, Delon I, Dolling H, Sanchis-Juan A, Shamardina O, Mégy K, Abbs S, Austin T, Bowdin S, Branco RG, Firth H; NIHR BioResource—Rare Disease; Next Generation Children Project, Rowitch DH, Raymond FL. Whole genome sequencing reveals that genetic conditions are frequent in intensively ill children. Intensive Care Med. 2019 May;45(5):627-636. doi: 10.1007/s00134-019-05552-x. Epub 2019 Mar 7. PMID: 30847515; PMCID: PMC6483967.