‘LIKE TRYING TO READ A MAP IN THE DARK’: UNDIAGNOSED GENETIC CONDITIONS, SERVICE USE AND FURTHER RESEARCH

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[1]Genetic Alliance UK is an alliance of over 200 patient organisations and the national charity working to improve the lives of patients and families affected by genetic conditions (www.geneticalliance.org.uk). SWAN UK, an initiative of Genetic Alliance UK, is UK-wide support network providing information and support to families of children affected by undiagnosed conditions.
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Background

- Recent advances in sequencing have identified the genetic basis of disease for 25-40% of patients (Sawyer *et al*, 2016), yet many remain undiagnosed.
- Evidencing the true scale and impact of undiagnosed genetic conditions is challenging, but essential if we are to meet the needs of thousands of families in the UK.

Aims

- To demonstrate the feasibility of collecting data on undiagnosed conditions.
- To highlight opportunities for further research and service improvement.

Method

The following data were reviewed:

- Snapshot of hospital admissions on 268 inpatients and coding data on 206 outpatients attending Birmingham Children’s Hospital (BCH) on Undiagnosed Children’s Day 2016.
- Qualitative data from families of children with undiagnosed genetic conditions across the UK via the SWAN UK public Facebook page and annual members’ survey.

What do we mean by undiagnosed genetic conditions?

The snapshot survey distinguished between those with and without a clear diagnosis at presentation to BCH. Some patients without a clear diagnosis may be suspected to have an extremely rare genetic condition which testing is not able to identify. These patients are described as having an **undiagnosed genetic condition** or a **‘syndrome without a name’ (SWAN).**

Key findings

The BCH Undiagnosed Day snapshot survey revealed:

1. A quarter of patients did not have a clear diagnosis at presentation to BCH. The proportions of such patients are large enough to suggest that the scale of their health need outweighs any single rare disease.

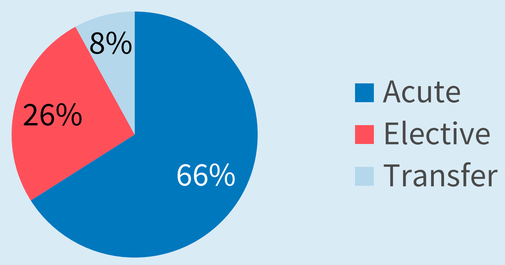
Table 1: Numbers of inpatients & outpatients with & without a clear diagnosis on presentation to BCH

	Outpatients	Inpatients	All patients
Clear diagnosis	139	218	357 (75%)
Without a clear diagnosis	67	50	117 (25%)
Total	206	268	474

2. Almost one third of the inpatients transferred from other hospitals did not have a clear diagnosis at the time of admission to BCH. The expertise of tertiary centres like BCH are crucial for establishing an underlying diagnosis and in managing and treating complex undiagnosed conditions.

3. Two thirds of inpatients without a clear diagnosis at presentation to BCH were acute admissions. Emergency Departments are a suboptimal route for families who have to summarise complex medical history to non-experts.

Chart 1: Percentage of inpatients without a clear diagnosis by type of admission



4. The majority of inpatients at presentation to BCH without a clear diagnosis had multiple health issues. Coordination of care is particularly important for patients affected by undiagnosed genetic conditions.

SWAN UK Facebook data revealed following challenges faced by families of patients affected by undiagnosed genetic conditions:

1. **Managing uncertainty**
2. **Getting timely access** to specialist care and support
3. **Communicating the child’s needs** without a diagnosis, especially in Emergency Departments
4. **Coordination of care**.

Next steps

- **Service audits** - to review numbers of patients without a diagnosis in other hospitals and build on findings of this study.
- **National surveillance** - to establish the national size of the patient population affected by undiagnosed genetic conditions in the UK.
- **Research** – to review the healthcare pathways that these patients use and conduct detailed long-term health economic studies to provide robust assessment of needs
- **Drive service improvement** - for those affected by undiagnosed genetic conditions.

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