Aetiological investigation of epilepsy in adults with a learning disability – A community audit

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Background

- The National Institute for Health and Care Excellence (NICE)[1] and International League Against Epilepsy (ILAE)[2] advise strenuous attempts to identify the aetiology of epilepsy as it often carries significant treatment implications.
- The ILAE divides aetiology into six categories selected because of their potential therapeutic consequences (structural; genetic; infectious; metabolic; immune; unknown).

Aims

- To demonstrate adherence to national standards for the aetiological investigation of epilepsy in the Epilepsy Service of Bromley Community Learning Disability Team (CLDT), Oxleas NHS Foundation Trust.

Methods

- Audit standards were derived from NICE[1] and ILAE[2].
- The key standard was:
  - 100% of patients should have sufficient aetiological investigation of their epilepsy, including a dysmorphism assessment, neuroimaging, and genomics, as appropriate.
- The population was defined as all patients open to the Epilepsy Service of Bromley CLDT in December 2021.
- Data was collected using a secure electronic database between December 2021 - January 2022.

Results

- 76 patients (52 male, 24 female) were audited with mean age 38 years (range 18-79 years).
- Learning disability severity included borderline (n=3), mild (n=28), moderate (n=24), severe (n=17) and profound (n=4).
- 59% (n=46) of patients had an identified epilepsy aetiology: 42% structural (n=32), 13% genetic (n=10), and 5% infectious (n=4). One patient had both genetic and infectious aetiology.
- 41% (n=31) of patients had unknown epilepsy aetiology, 58% (n=18) of whom had outstanding investigations – see figure 1.
- In 72% (n=13) of patients there was no apparent explanation for investigations to be outstanding.
- Genomics was the commonest outstanding investigation (70%, n=14), followed by neuroimaging (20%, n=4) and dysmorphism assessment (10%, n=2) – see figure 2.
- In addition, 40% (n=12) of patients were newly eligible for whole genome sequencing – a recent development within 12 months and thus excluded from outstanding investigations.

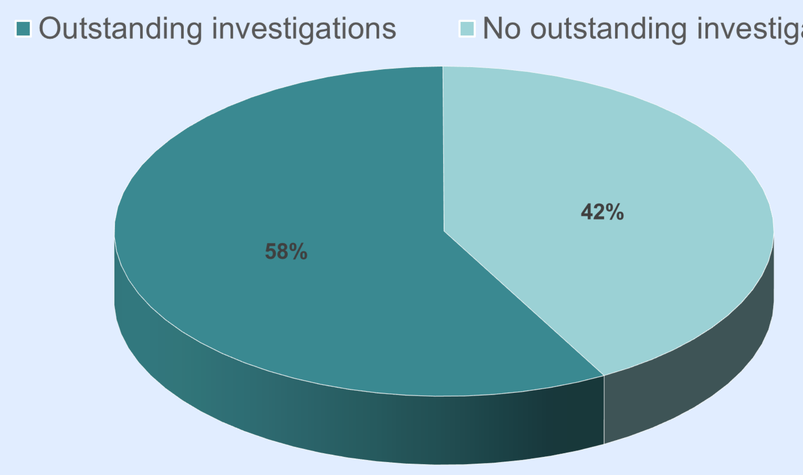

Figure 1 – Percentage of patients with unknown epilepsy aetiology who had outstanding investigations.

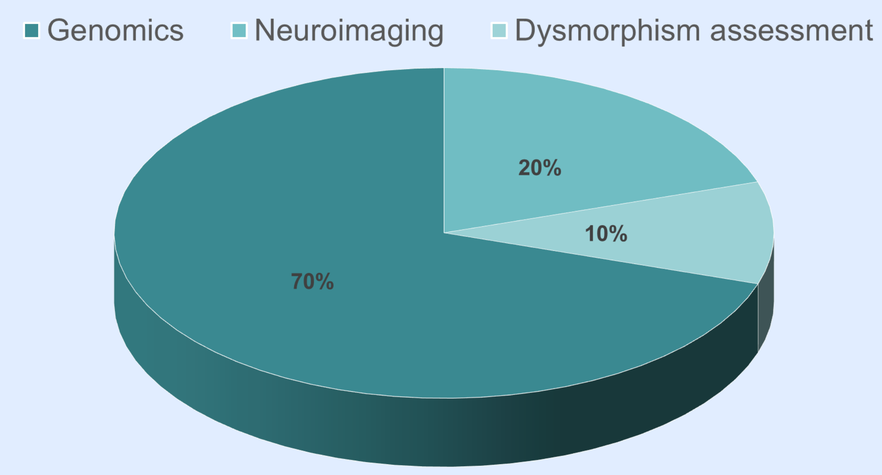

Figure 2 – Percentage of patients with outstanding genomic, neuroimaging and dysmorphism investigations.

Discussion

- Over half of patients with unknown epilepsy aetiology had outstanding investigations – the majority with no apparent explanation.
- Genomics was the commonest outstanding investigation, and the new availability of whole genome sequencing will likely compound this. Clinician training on genomic testing indications and practicalities is therefore required.
- An epilepsy service protocol could help standardise the local approach to the aetiological investigation of epilepsy.

Conclusion

- This audit demonstrates adherence to national standards is below 100%.
- Where appropriate, patients with outstanding investigations will be approached.
- Dissemination of findings and an action plan are required before re-audit.

References

1. National Institute for Health and Care Excellence (2012) Epilepsies: diagnosis and management [CG137]. Access: https://www.nice.org.uk/guidance/cg137.
2. International League Against Epilepsy (2017) ILAE classification of the epilepsies: Position paper of the ILAE Commission for Classification and Terminology. Access: https://doi.org/10.1111/epi.13709.

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