Tertiary Centre Experience In Investigating And Diagnosing Floppy Babies

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INTRODUCTION

- Floppy babies are one of the commonest neonatal referrals to Paediatric Neurologists.
- Clinical examination is an essential first step in the diagnostic journey.
- With the advent of new time-critical, treatments for neurological conditions, e.g. gene therapy for SMA, an effective pathway for investigation is essential to diagnose the aetiology.

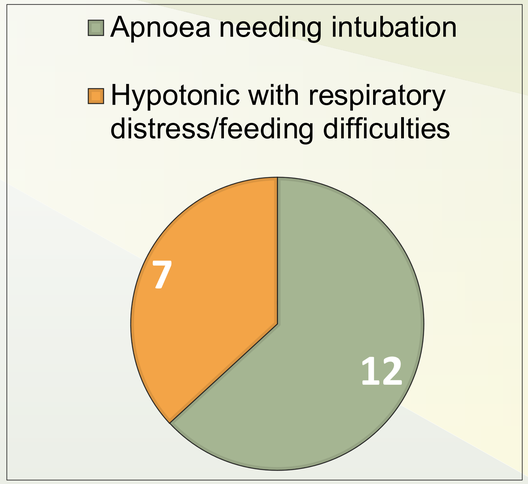

Figure 1. Presenting Symptoms

METHODS

- A retrospective review of all floppy babies admitted to NICU/PICU at Addenbrookes hospital, Cambridge between 2017-2020.

RESULTS

- Of the 19 babies, 13 were male and 6 female.

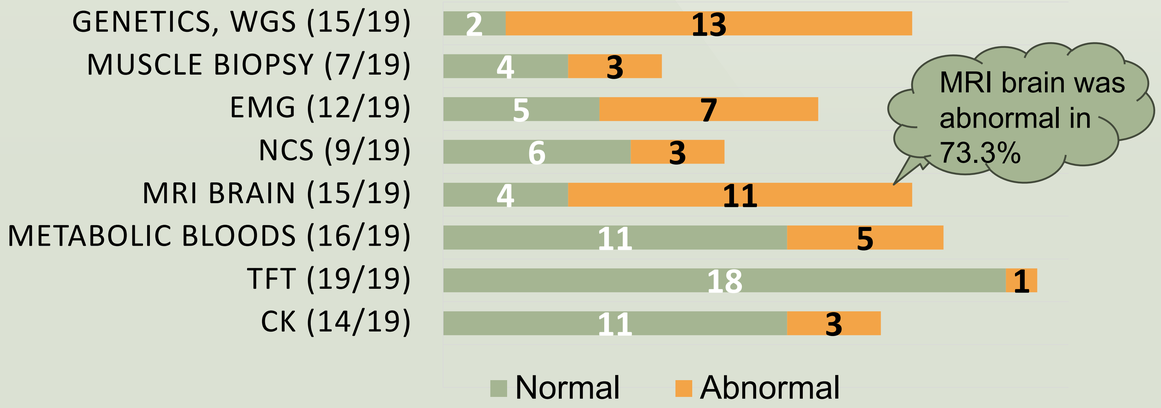

Figure 2. Investigations of babies and the respective outcomes.
WGS – Whole Genome Sequencing, EMG/NCS – Electromyography/ Nerve Conduction Studies, TFT – Thyroid Function Test, CK – Creatinine Kinase

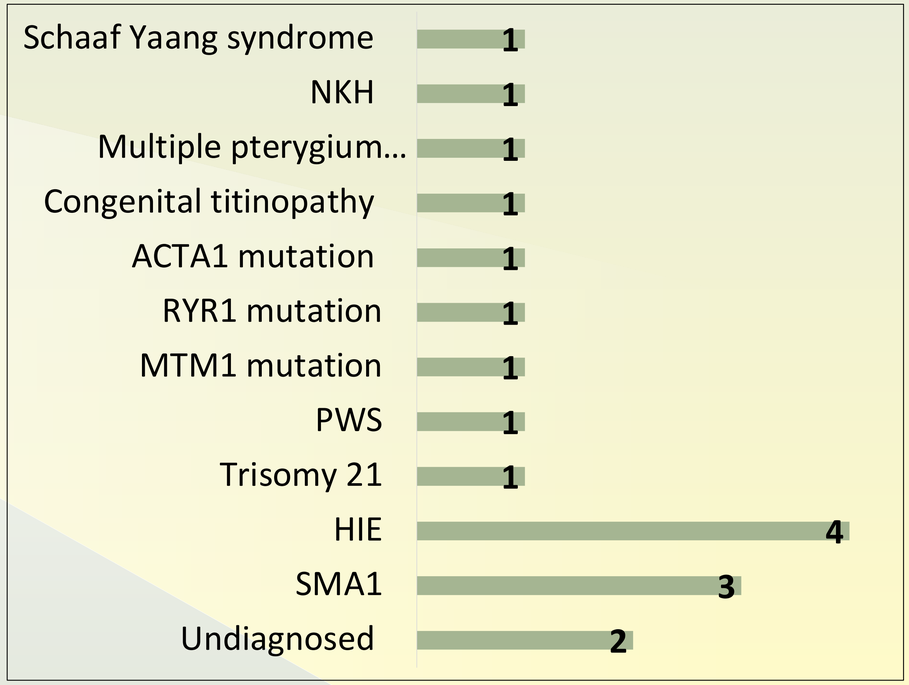

Figure 3. Diagnosis of our cohort. NKH – Non Ketotic Hyperglycinemia, PWS – Prader Willi Syndrome, HIE – Hypoxic Ischemic Encephalopathy, SMA – Spinal Muscular Atrophy

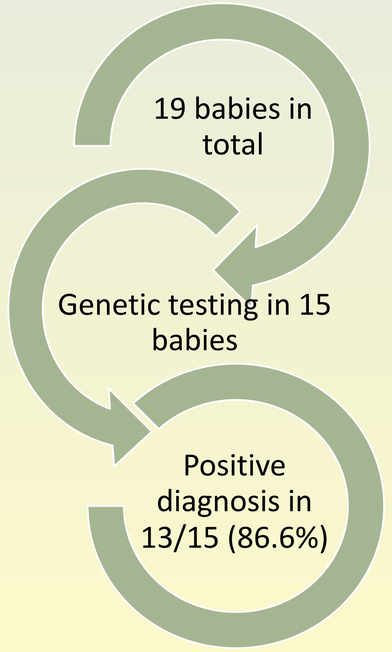

Figure 4. Results of Genetic testing

CONCLUSIONS

- With the changing landscape of treatment availability, it is imperative that we use rapid genetic testing to diagnose conditions where there are known treatments.
- Advances in trio WGS, have reduced the need for invasive investigations like lumbar-puncture, EMG, NCS and muscle biopsy.
- This study highlights the need for using genetic testing as first line investigation for floppy babies and ensure it is reflected in the local-guidelines.

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