Trajectory Of Project Baby Bear Rapid Genome Sequencing Project: A Review of Long-Term Follow Up at a Single Center

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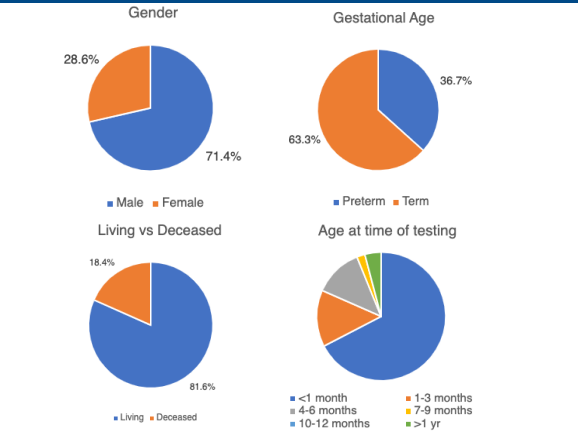
Introduction

- Project Baby Bear was a prospective quality improvement project which demonstrated the clinical utility and economic impact of ultrarapid or rapid whole genome sequencing (rWGS) in neonatal and pediatric intensive care units for Medi-Cal beneficiaries
- This study found that rWGS provided a diagnosis for 40% of the patients and changed medical care for 32% of them.
- However, there is limited information on the long-term clinical outcomes and follow up of these individuals.

Methods

- A retrospective chart review of 49 participants was conducted using data from one large, tertiary academic center involved in Project Baby Bear

Demographics



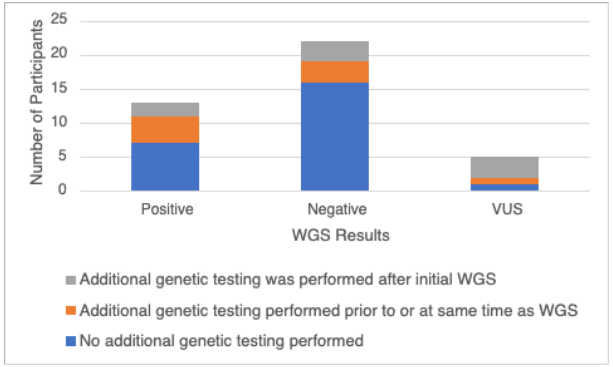


Figure 1. Whole genome sequencing results

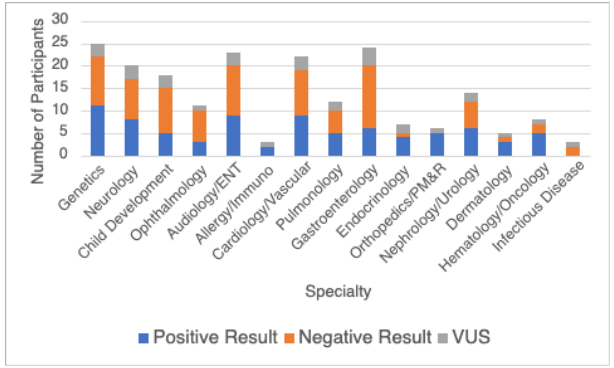


Figure 2. Specialty care follow-up

Results

- WGS results were as follows: 32% pathogenic or likely pathogenic, 55% negative, and 12.5% variant of uncertain significance (Figure 1).
- No additional genetic testing was pursued in a total of 60% of individuals. 20% had additional genetic testing performed prior to or at the same time as WGS and the other 20% had additional genetic testing pursued after initial WGS. (Figure 1)
- Most patients had at least one follow-up specialty visit at UC Davis Health or an outside health system. The specialty distribution is seen in Figure 2.

Discussion

- Only 60% of infants who received WGS were seen by Genetics for follow-up and further genetic testing was conducted in 20%.
- The initial WGS analysis was phenotypically driven, but the clinical phenotype can evolve rapidly and change over time.
- Improved knowledge and technology over time would also likely help identify underlying etiology with re-analysis of the genome.

Conclusion

- From review of the long term follow up for our cohort, we found that we are not capturing all individuals who received WGS through Project Baby Bear into follow up.
- Future directions for this project include contacting families prospectively for additional patient-centered perspectives and further chart review of the referral process to genetics outpatient clinic following a hospital admission.

Acknowledgements

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