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Capillary blood microsampling for genome sequencing in healthy newborns

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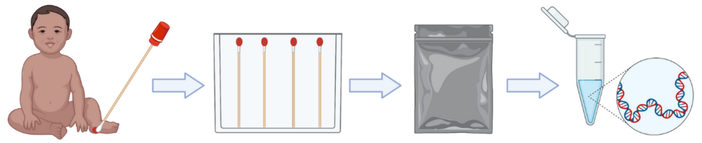
Background

- There is growing interest in expanded newborn screening (NBS) using genome sequencing (GS).
- However, it is a challenge to obtain DNA from healthy newborns.
- Standard NBS dried blood spots are not routinely available for additional testing and do not consistently yield sufficient DNA.
- Infant buccal/saliva samples are difficult to collect, and venipuncture is not well accepted by parents.
- We explored whether microsampling is acceptable to new parents and could yield sufficient DNA for GS.

Methods

- A research assistant approached families in the Brigham and Women's Hospital well-baby nursery in Boston, Massachusetts, and informed consent was obtained.
- A research nurse performed a single heel stick and collected blood using a commercial microsampling kit.
- Parents completed a brief online survey after collection.

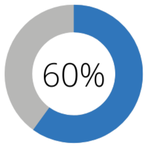
Microsampling Workflow



Heel stick | Volumetric absorptive microsampling | Temperature-stable shipping | DNA extraction

Approach and Sample Collection

- The main reasons for declining participation were:
  - Scheduling (n=4)
  - Opposition to extra heel stick (n=2)
- 9 families completed a follow-up survey after collection.
  - All families reported that their babies experienced "no pain" or "some pain" from the heel stick.
- Parents expressed that heel warmers made collection more comfortable for infants.

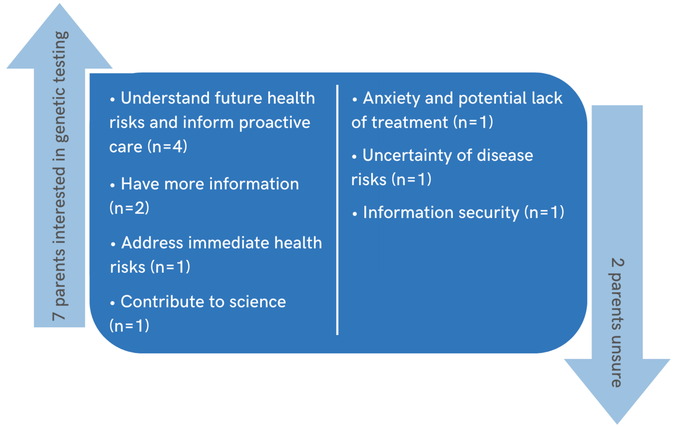


Of 20 families approached and oriented to the study, 12 (60%) of families consented to a heel stick.

All microsampling extractions have yielded sufficient DNA for genome sequencing (average 1.8 micrograms).

Attitudes Towards Newborn Genetic Testing

- Of the 12 participating families, 9 families completed a follow-up survey.
- Parents equally preferred less invasive samples including heel stick microsampling, saliva, or cheek swab over venipuncture.
- Parents reported their interest in newborn genetic testing and their reasons for interest.



Participant Experience

What are the main reasons you would be interested in genetic testing?

> *More information is beneficial.*
> *If there was an actionable finding.*
> *To perform any preventative or care needed.*
> *To learn more about my baby.*

What are the main reasons you would not be interested in genetic testing?

> *I don't want to structure my child's life around fear of possibilities.*
> *Concerns about security of genetic info.*

Conclusions

- This pilot study suggests it is feasible to collect an additional heel stick from healthy newborns, and that capillary blood microsampling can yield sufficient DNA for GS.
- Parents found this method acceptable, and the majority of those enrolled expressed interest in genetic testing to inform proactive care.

Future Directions

- We continue to validate this sample type in infants <6 months old at Boston Children's Hospital.
- The BabySeq Project will attempt to leverage this sample type for an expanded randomized controlled trial of GS in healthy infants.

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