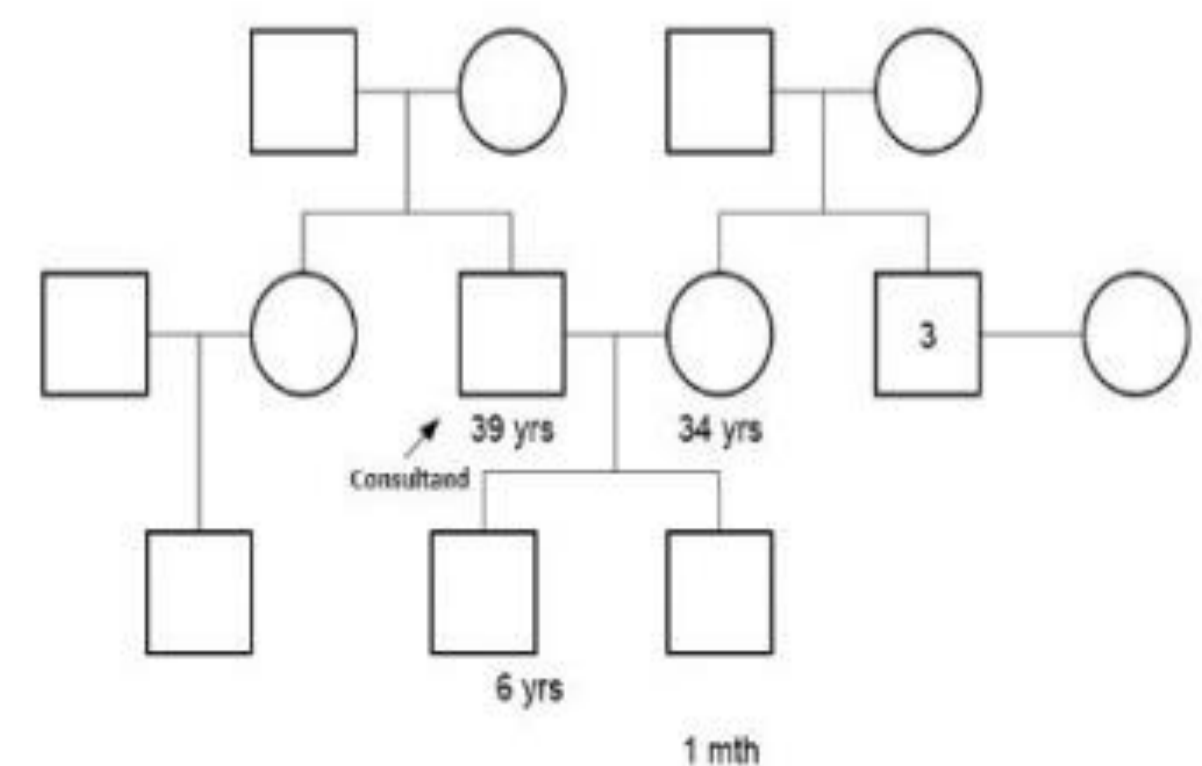


Introduction

- Direct-to-consumer (DTC) genetic testing services have revolutionized access to personal genetic information, empowering individuals to explore their ancestry, health predispositions, and carrier status from the comfort of their homes.
- However, the complexity of genetic data interpretation and the potential psychological impact of unexpected findings underscore the critical role of genetic counselling in these services
- Genetic counsellors serve as integral intermediaries, translating complex genetic information into actionable insights, assisting individuals in navigating medical management options, and addressing ethical, legal, and social implications.
- Through collaborative partnerships with DTC companies, genetic counsellors uphold standards of care, ensuring that consumers receive accurate, comprehensive, and culturally sensitive genetic counselling services

Case details and Methodology

Asymptomatic 1-month-old male child, with no complaints. Stem cells from cord blood for this child were stored, and directly underwent an NGS based Whole Exome Sequencing for monogenic disorder



Result

Gene	Variant	Zygoty	Disease	Inheritance
<i>LAMB2</i>	c.4573C>T (p.Gla1525*)	Heterozygous	Nephrotic syndrome, type 5	Autosomal Recessive (AR)
<i>FAH</i>	c.192G>T (p.Gin6His)	Heterozygous	Tyrosinemia, type 1	

Counselling and Challenges

- Baby's father was alarmed upon seeing the report and sought genetic consultation, believing it meant the child had the disorders. He was then explained the result means his child is a carrier and not affected by these conditions as they follow an AR pattern of inheritance.
- If the individual has both altered copies, then it will lead to an AR condition. The father revealed that he was unaware of what to expect from the genetic test report leading to this anxiety.

Discussion

- In the era of DTC genetic testing, genetic counselling remains indispensable to avoid unnecessary anxiety due to misunderstanding of the results and making inappropriate health decisions.
- Direct-to-consumer (DTC) genetic testing has revolutionized access to personal genetic information, allowing individuals to explore health predispositions and carrier status.
- However, the complexity of genetic data interpretation and potential psychological impacts highlight the crucial role of genetic counselling to act as intermediaries in translating complex genetic data into actionable insights, guiding medical management options, and addressing ethical, legal, and social implications.

Conclusion

- Many individuals opt for DTC GT, and view the results as valid and potentially impactful for their healthcare, and want more thorough explanations in "layman's terms," a pooling of their results with their family and personal health history and a "game plan" - for which they may seek genetic counselling.
- Nowadays, everyone has access to online resources, including resources typically used by genetics clinicians. These resources help them with:

Effective contracting when starting the clinic visit, especially determining motivations for genetic counseling, results that are concerning/confusing and resources already accessed

Ascertainment and management of expectations and clearly communicating if and why all results may not be reviewed;

Explaining how DTC GT differs from clinical genetic testing and why additional testing may not be indicated

Listening to and not dismissing any patient concerns about their results.

- Currently scientific evidence are limited to offer genetic test for asymptomatic new born babies without family history or clinical indication which can create complex psychological implications on parents
- Pre Test counseling should offer given it plays a significant role.
- For individuals who seek genetic counselling about DTC GT results, the pre and post test counselling can help inform case preparation and provision of genetic counselling. Hence, Collaborative partnerships with DTC companies ensure accurate, comprehensive, and culturally sensitive genetic counselling services.

Reference

1. Marzulla, T., Roberts, J. S., DeVries, R., Koeller, D. R., Green, R. C., & Uhlmann, W. R. (2020). Genetic counseling following direct-to consumer genetic testing: Consumer perspectives. *Journal of Genetic Counseling*. doi:10.1002/jgc4.1309

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