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Response to Correspondence on "Genomic testing for suspected monogenic kidney disease in children and adults: a health economic evaluation" (Lombardi and Mesnard, 2023)

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Response

We write to acknowledge the recent correspondence by Lombardi and Mesnard (2023) and thank them for their positive comments regarding the quality, importance and novelty of our work.[1] We concur that economic evaluation studies from a wide variety of jurisdictions and health systems are very much needed.

We also agree that real-world data have a crucial role to play in understanding and valuing the health economic impacts of genomics and precision medicine for both probands and family members and support evidence-based regulatory and reimbursement decision-making. It is critical that relevant data infrastructure and systems are set up to support the collection and use of real-world evidence for informing healthcare priorities.

Beyond considering the source of healthcare utilization data, we would also like to highlight the importance of capturing the broader 'utility' of genomics[2] to reflect the diagnostic, personal and clinical value to patients and families. While our work has attempted to address this,[1] more economic evaluations incorporating spillover effects to families[3] and valuations of the broader genomic utility[4] are needed to support the sustainable and equitable translation of genomics into clinical care.

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