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

You Wu, MPH[1,2,3], Kushani Jayasinghe, MBBS[3,4,5,6], Zornitza Stark, BMBCh DM[2,7,8], Catherine Quinlan, MD(Res)[3,6,8,9], Chirag Patel, MD[6,10], Hugh McCarthy, PhD[11,12,13], Amali C. Mallawaarachchi, PhD[14,15], Peter G. Kerr, PhD[4,5], Stephen I Alexander, MD[11,12,13], Andrew J. Mallett, PhD[6,16,17,18,*], Ilias Goranitis, PhD[1,2,3,6,*] on behalf of The KidGen Collaborative investigators

[1] Health Economics Unit, Centre for Health Policy, Melbourne School of Population and Global Health, University of Melbourne, Melbourne, VIC, Australia

[2] Australian Genomics Health Alliance, Melbourne, VIC, Australia

[3] Murdoch Children's Research Institute, Melbourne, VIC, Australia

[4] Department of Nephrology, Monash Medical Centre, Melbourne, Australia;

[5] Monash University, Melbourne, Australia;

[6] The KidGen Collaborative, Australian Genomics Health Alliance, Melbourne, Australia

[7] Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Melbourne, Australia

[8] Department of Pediatrics, University of Melbourne, Melbourne, Australia

[9] Department of Pediatric Nephrology, Royal Children's Hospital, Melbourne, Australia

[10] Genetic Health Queensland, Royal Brisbane and Women's Hospital, Brisbane, Australia;

[11] Centre for Kidney Research, Children's Hospital at Westmead, Sydney, Australia;

[12] Sydney Children's Hospitals Network, Sydney, Australia;

[13] Faculty of Medicine, The University of Sydney, Sydney, Australia;

[14] Department of Medical Genetics, Royal Prince Alfred Hospital, Sydney, Australia;

[15] Garvan Institute of Medical Research, Sydney, Australia;

[16] Institute for Molecular Bioscience and Faculty of Medicine, The University of Queensland, Brisbane, Australia

[17] Department of Renal Medicine, Townsville University Hospital, Townsville, Australia

[18] College of Medicine & Dentistry, James Cook University, Townsville, Australia

* Joint Senior Co-Authors. Contact details: Email: ilias.goranitis@unimelb.edu.au; Telephone: +61 3 8344 9959; Email: andrew.mallett@health.qld.gov.au; Telephone: +61 7 4433 5091

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.

References

1. Wu Y, Jayasinghe K, Stark Z, et al. Genomic testing for suspected monogenic kidney disease in children and adults: a health economic evaluation. Genet Med. 2023:100942.
2. Mallett A, Stark Z, Fehlberg Z, Best S, Goranitis I. Determining the utility of diagnostic genomics: a conceptual framework. Hum Genomics. 2023;17(1):75.
3. Wu Y, Al-Janabi H, Mallett A, et al. Parental health spillover effects of paediatric rare genetic conditions. Qual Life Res. 2020;29(9):2445-2454.
4. Goranitis I, Best S, Christodoulou J, Stark Z, Boughtwood T. The personal utility and uptake of genomic sequencing in pediatric and adult conditions: eliciting societal preferences with three discrete choice experiments. Genet Med. 2020;22(8):1311-1319.