In reply: We thank Martin and colleagues[1] for their critical appraisal of our review on genetic testing in cardiovascular disease, published in the *MJA*.[2] We agree that the utility of genetic testing needs to consider the burden of disease, the age of onset, and treatment options available to individuals identified with the causal genetic variant. We also concur with Martin and colleagues regarding the value of early detection and treatment of familial hypercholesterolaemia.[2] We particularly appreciate the emphasis of familial hypercholesterolaemia being a disorder frequently identified in paediatric patients, and the proposed clinical pathway for prevention of atherosclerosis and myocardial infarction, with consideration of lipid-lowering treatment after maximal lifestyle interventions from age six for those with homozygosity.[3]

In the general population, current expert consensus guidelines continue to recommend genetic testing as a confirmatory tool following identification using clinical tools such as Simon Broome Diagnostic Criteria or the Dutch Lipid Clinic Network Score.[3,4] However, identification of a familial hypercholesterolaemia-associated variant in an individual justifies further cascade variant testing in first-, second-, and even third-degree biological relatives for earlier diagnosis and intervention.[5,6]

As with many conditions highlighted in our article, the role of genetic testing in the identification of familial hypercholesterolaemia continues to evolve with improved understanding of the disease genetic architecture, clinical experience incorporating genomic testing, and access to sequencing technologies. Health economics, guideline development, and policy changes will be key to maximising the value of all genetic tests in the cardiovascular disease space.

Michael P Gray[1,2]
Gemma A Figtree[1,2]

1 University of Sydney, Sydney, NSW.
2 Kolling Institute of Medical Research, University of Sydney, Sydney, NSW.

michael.gray@sydney.edu.au

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executive committee member for CPC Clinical Research, founding director and CMO for Prokardia and Kardiomics, and executive committee member for the CAD Frontiers A2D2 Consortium. In addition, GAF serves as CMO for the non-profit, CAD Frontiers, with industry partners including Novartis, Amgen, Siemens Healthineers, ELUCID, Foresite Labs LLC, HeartFlow, Canon, Cleerly, Caristo, Genetech, Artyra, and Bitterroot Bio and Allelica. In addition, GAF has the following patents: "Patent Biomarkers and Oxidative Stress" awarded USA May 2017 (US9638699B2) issued to Northern Sydney Local Health District, "Use of P2X7R antagonists in cardiovascular disease" PCT/AU2018/050905 licensed to Prokardia, "Methods for treatment and prevention of vascular disease" PCT/AU2015/000548 issued to the University of Sydney/Northern Sydney Local Health District, "Wound healing methods" PCT/AU2022/050129 issued to the University of Sydney, "Wound healing compositions" PCT/AU2022/050130 issued to the University of Sydney, "Methods for predicting coronary artery disease" AU202290266 issued to the University of Sydney, and the patent "Novel P2X7 Receptor Antagonists" PCT/AU2022/051400 (23.11.2022), International App No: WO/2023/092175 (01.06.2023), issued to the University of Sydney. ■

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