

Functional Evidence Evaluation

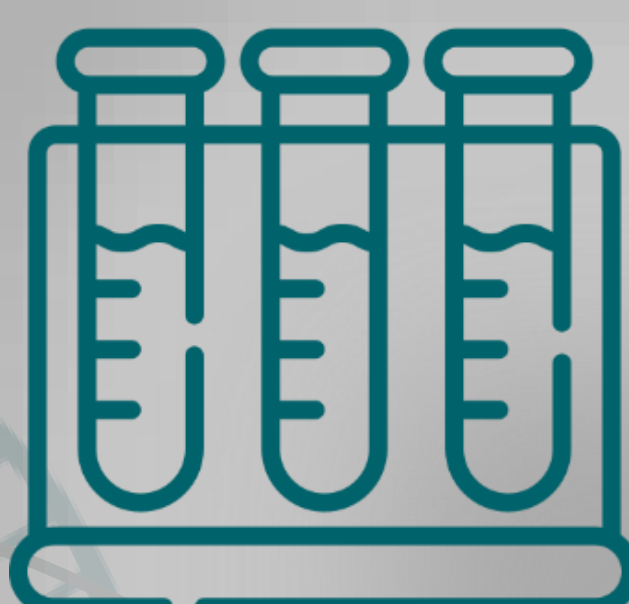
RESOLVING UNCERTAINTY IN VARIANT CLASSIFICATION BY INCREASING THE APPLICATION OF EXPERIMENTAL DATA IN CLINICAL DIAGNOSTICS

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by the AusMAVE Education Project

Our mission



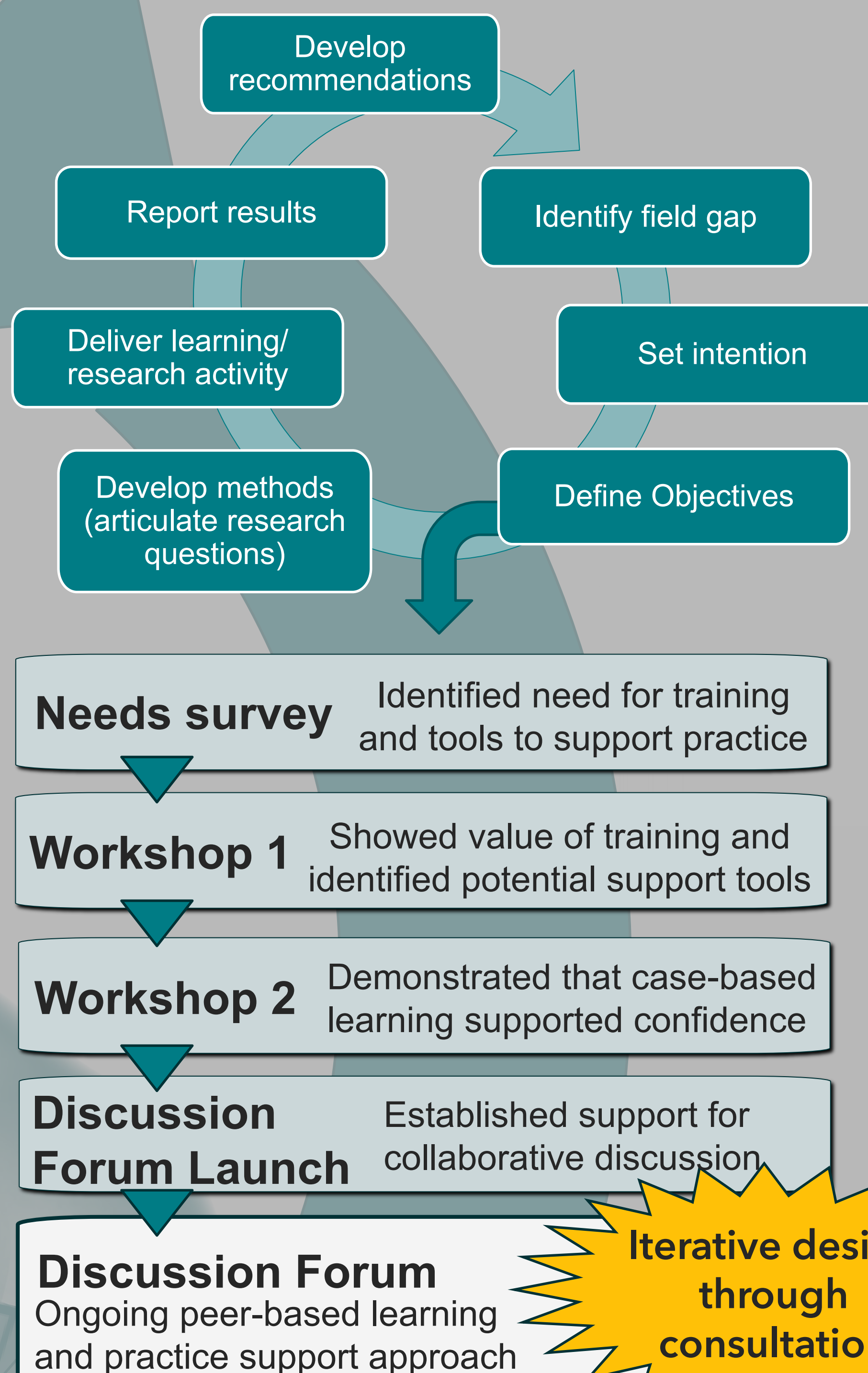
Experimental data can be applied as **functional evidence** in variant curation if appropriately evaluated, and yet functional evidence is **under-represented** in curation.

High throughput experimental assays such as Multiplexed Assays of Variant Effect (MAVEs) are **rapidly increasing** so the **gap** in functional evidence application will continue to expand.



We aim to **improve the use of functional evidence** in genomic diagnostic testing or many patients will remain undiagnosed unnecessarily.

Defining the problem



Campaign for better practice

Delivering a community forum discussing assays and practice

- Shared resources & public events
- Community review of tools and assays
- Participate in activities supporting new resources & recommendations



DISCUSSION FORUM!!

image: Flaticon.com

ONGOING

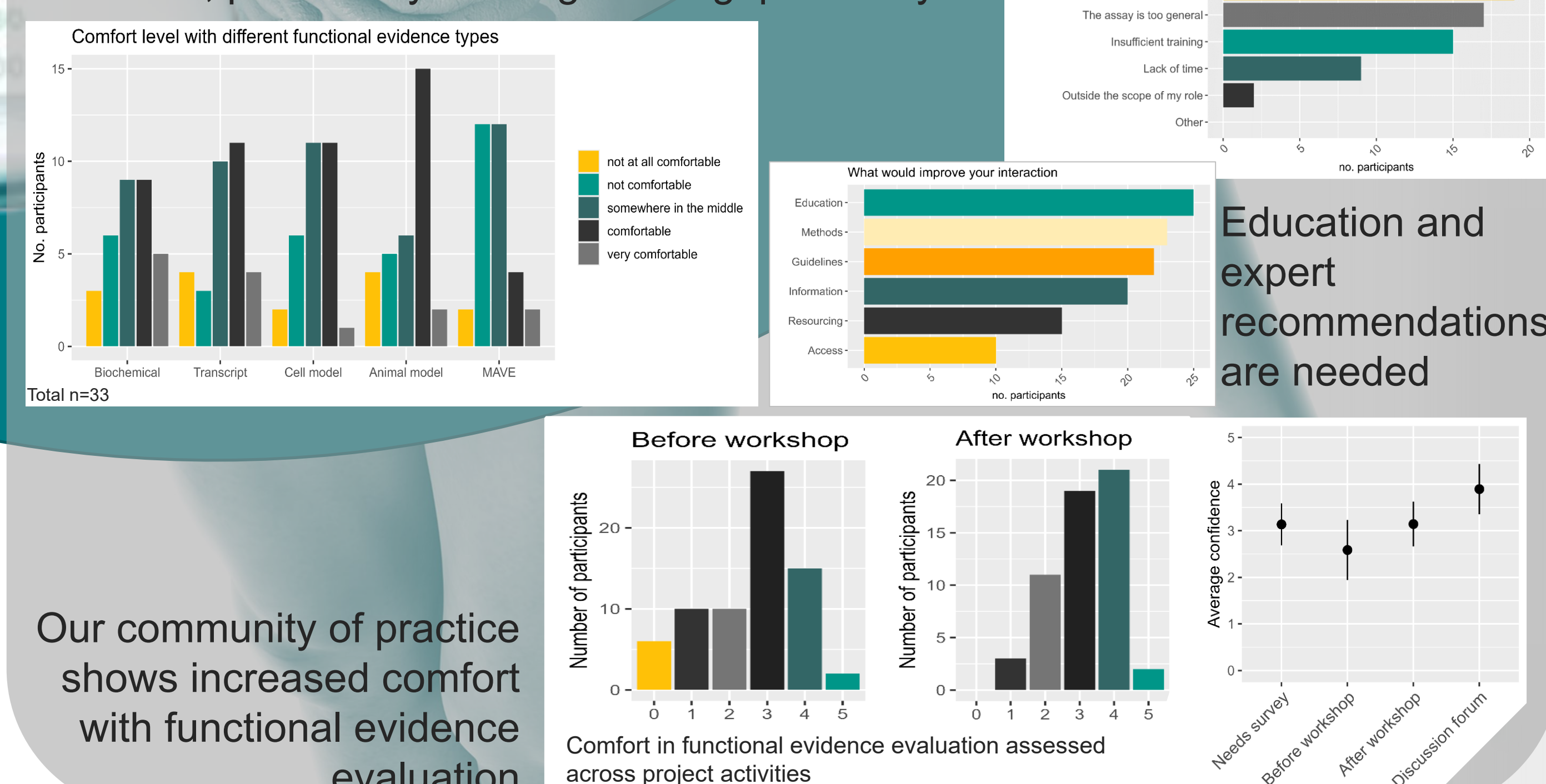
Online resources



For more details go to www.qimrb.edu.au/ausmave, Villani et al 2025 PMID: 40480201 & doi.org/10.1101/2025.11.10.25339935

Building solutions

Curators have variable comfort in functional evidence evaluation, particularly with high-throughput assays



Acknowledgements



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