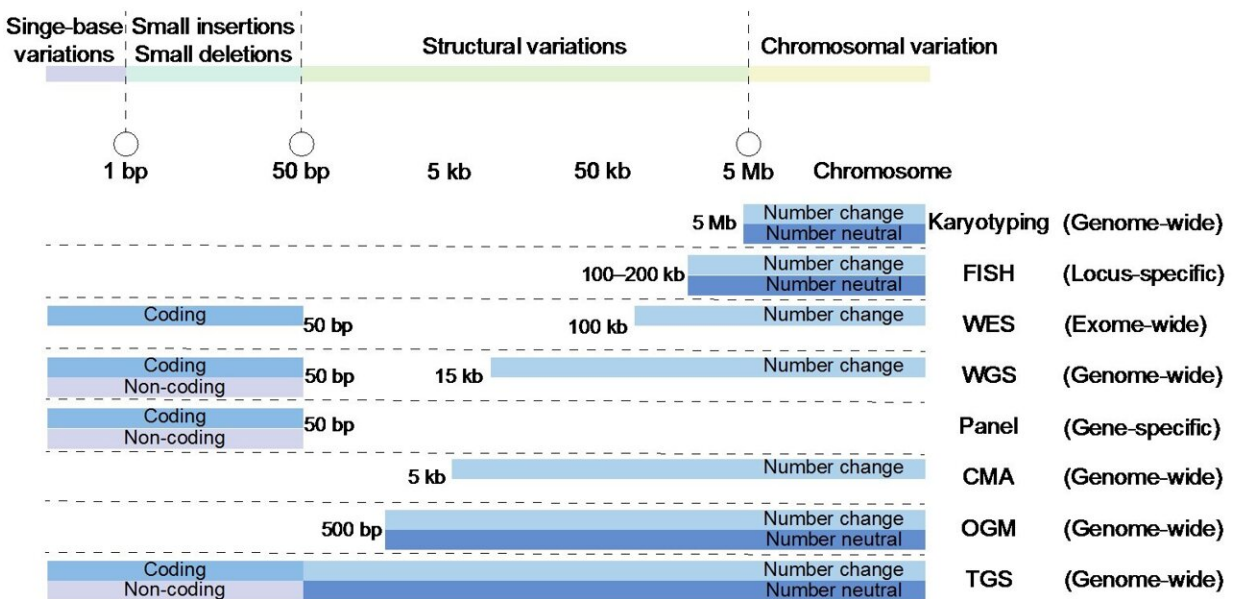


Progress in genetic testing in congenital heart disease

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The coverage of variants yielded by testing technologies is indicated. Structural variants may entail duplications, deletions, and unbalanced insertions, while positional changes may involve balanced translocations, insertions, or inversions. Credit: Science China Press

Congenital heart disease (CHD) includes a variety of structural malformations affecting the heart and blood vessels. With growing understanding of the molecular processes involved in heart development, genetic variants have been proven to play a crucial role in the

development of CHD.

Researchers have identified a range of genetic variations associated with both syndromic and non-syndromic CHD, including single nucleotide variations, structural variations, and chromosomal abnormalities. The research is [published](#) in the journal *Medicine Plus*.

As [genetic testing](#) technologies improve, the application of genetic testing in CHD patients yields valuable insights in the aspects of both clinical and research settings.

With the advancement of cohort studies, genetic data can be effectively collected from genetic testing and categorized by pathogenicity, enhancing comprehension of CHD genetics. Many tools have been created to interpret variants identified in both coding and non-coding regions, providing valuable information for [clinical practice](#) in CHD management. The integration of genetic testing could benefit CHD management in several aspects, including diagnosis, prognosis prediction, and recurrence risk estimation.

In conclusion, with ongoing advancement in understanding of genetic testing and CHD, [health care providers](#) can effectively incorporate genetic testing into the broader framework of CHD care, leading to better patient outcomes.

More information: Han Gao et al, Progresses in genetic testing in congenital heart disease, *Medicine Plus* (2024). [DOI: 10.1016/j.medp.2024.100028](#)

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