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Integrating CMR and Genetics to Improve ATTR-CM Diagnosis

Ryan Quigley:

Welcome to *AudioAbstracts* on ReachMD. I'm Ryan Quigley, and today, I'll be highlighting an April 2025 review from *Current Cardiology Reports* exploring how the integration of cardiac magnetic resonance, or CMR, imaging and genetic data is advancing our knowledge of cardiomyopathy pathogenesis while improving diagnostic and prognostic capabilities. This review covers a broad spectrum of genetic cardiomyopathies, but for today's discussion, I'll be focusing on hereditary transthyretin amyloidosis with cardiomyopathy, or ATTR-CM.

In hereditary ATTR-CM, an autosomal-dominant variant in the *TTR* gene leads to the deposition of misfolded transthyretin protein fibrils within the myocardium. These extracellular deposits lead to progressive myocardial infiltration, which can be identified using advanced CMR techniques.

The review highlights several key imaging modalities for detecting ATTR infiltration. Late gadolinium enhancement, or LGE for short, native T1 mapping, and extracellular volume, or ECV, quantification are particularly valuable.

One of the distinctive imaging hallmarks is the reverse nulling pattern observed on post-contrast inversion recovery sequences. In this setting, gadolinium clears from the blood pool more rapidly than from the amyloid-laden myocardium. This inversion of the expected signal behavior serves as a specific indicator of cardiac amyloidosis. What's more, ATTR-CM is typically characterized by subendocardial or transmural LGE with increased native T1 and ECV values, reflecting myocardial amyloid infiltration.

The authors also mention the growing role of quantitative perfusion imaging in ATTR-CM. Patients demonstrate markedly reduced stress myocardial blood flow and impaired myocardial perfusion reserve, consistent with amyloid-related microvascular dysfunction. These findings correlate with myocardial involvement and disease severity, supporting the potential value of perfusion CMR in disease assessment.

Beyond diagnosis, these imaging features carry prognostic significance. Elevated ECV and native T1 values have been shown to correlate with higher amyloid burden and increased mortality risk.

Ultimately, integrating CMR imaging with genetic data may refine disease classification, strengthen risk stratification, and support future precision-based therapies. At the same time, the authors note that widespread implementation is limited by the need for specialized expertise, highlighting radiomics and artificial intelligence as promising tools to streamline CMR analysis and improve clinical efficiency.

This has been an *AudioAbstract*, and I'm Ryan Quigley. To access this and other episodes in our series, visit ReachMD.com, where you can Be Part of the Knowledge. Thanks for listening!

Reference

Paudel B, Pan J, Singulane CC, et al. Cardiac Magnetic Resonance Guidance for the Pathogenetic Definition of Cardiomyopathies. *Curr Cardiol Rep*. 2025;27(1):85. Published 2025 Apr 16. doi:10.1007/s11886-025-02233-8