THE ROLE OF TENASCIN-X IN IDIOPATHIC GASTROPARESIS: WGS ANALYSIS

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Whole genome sequencing of over 1700 gastroparesis patients reveals statistically significant enrichment of moderate to rare TNX gene variants, extending beyond confirmed Ehlers-Danlos syndrome cases to the broader idiopathic gastroparesis population.

KEY POINTS

- The study identified 83 non-synonymous TNX variants with at least 2-fold enrichment in gastroparesis patients, with minor allele frequency less than 0.05, several of which have been previously reported in association with Ehlers-Danlos syndrome in CLINVAR.
- TNX enrichment was observed not only in patients with confirmed Ehlers-Danlos syndrome status but across the entire idiopathic gastroparesis cohort, suggesting a broader role in gastroparesis pathophysiology.
- One specific variant, TNX:NM_019105:exon26:c.A9044G:p.K3015R with CADD score of 23.3, was identified in two patients with Ehlers-Danlos syndrome and idiopathic gastroparesis, and TNX microdeletions were also detected in a subset of patients.
- TNX is a glycoprotein that regulates tissue structure through anti-adhesive interactions with collagen; TNX-deficient mice exhibit accelerated gastric emptying and increased vagal afferent responses to gastric distension that correlate with gastrointestinal symptoms in TNX-deficient patients.
- The authors note this finding would not be delineated with classic genome-wide association study approaches and all authors are employees of Vanda Pharmaceuticals Inc.

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