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CMT-aaRS variants cause translational defect alleviated by cognate tRNA enhancement

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Dominant missense mutations in eight aminoacyl-tRNA synthetases (aaRSs) are associated with axonal and intermediate forms of Charcot-Marie-Tooth (CMT) disease. The molecular mechanisms by which these ubiquitously expressed enzymes cause selective peripheral neuronal degeneration remain elusive. To address this, we use cell models and patient-derived materials. Employing a variety of high-resolution approaches (e.g. microscopy, NGS, AI-based, biochemical methods), we discovered a common mechanism of tRNA sequestration that alters translation and is likely linked to accelerated neuronal degeneration.

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