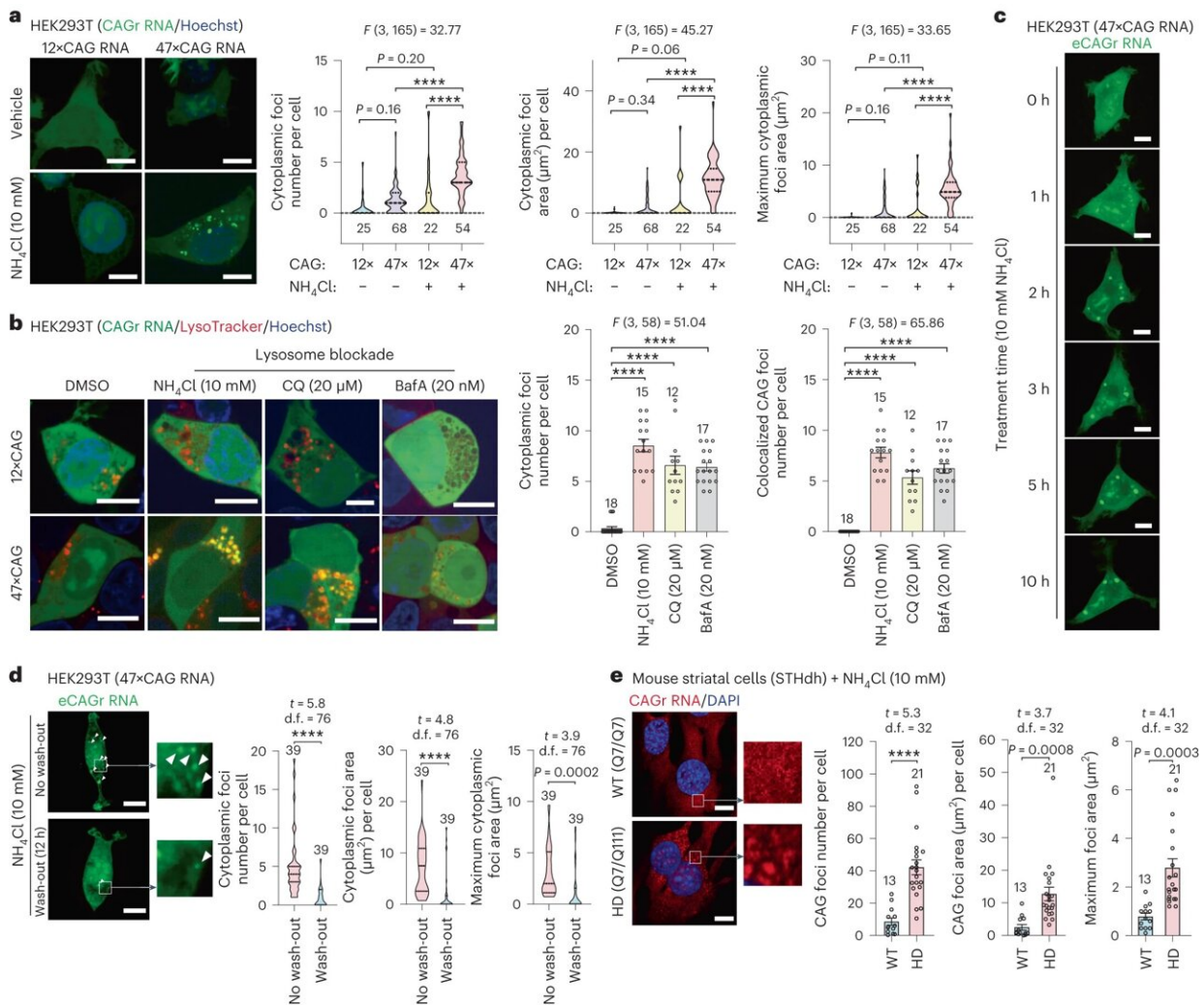


Study uncovers potential new source of genetic mutations that cause neurodegenerative disease

August 17 2023



The eCAGr RNA forms cytoplasmic gel-like condensates degraded by the lysosomes. **a**, Representative images and quantifications of eCAGr RNA

condensates (foci) in transfected HEK293T cells expressing the indicated RNA together with MS2CP–YFP as the foci detector. **b**, Representative images and quantifications of eCAGr RNA foci and the lysosomes in HEK293T cells treated with the indicated lysosome inhibitors (NH_4Cl , chloroquine (CQ) or bafilomycin A1 (BafA)). **c,d**, Snapshots and quantifications of the representative live-cell imaging (from 20) showing that the eCAGr RNA foci appeared in the cytoplasm after adding the lysosome inhibitor NH_4Cl but not the vehicle (culture medium) control (**c**), and disappeared after washing out NH_4Cl (**d**). eCAGr RNA foci are indicated by white arrows (**d**). **e**, Representative images and quantifications of RNA foci by RNA FISH in HD STHdh cells treated with the indicated lysosome inhibitors versus WT STHdh cells. Mean \pm s.e.m.; one-way ANOVA with multiple comparisons (**a** and **b**), or unpaired two-tailed *t*-tests (**d** and **e**). *n* = number of cells examined over three independent experiments. *****P*

Citation: Study uncovers potential new source of genetic mutations that cause neurodegenerative disease (2023, August 17) retrieved 29 April 2024 from <https://medicalxpress.com/news/2023-08-uncovers-potential-source-genetic-mutations.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.