

# Delineating the Molecular Spectrum and the Clinical, Imaging and Neuronal Phenotype of Chopra-Amiel-Gordon Syndrome

NCT05528744

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**Status** RECRUITING  
**Sponsor** Boston Children's Hospital  
**Enrollment** 125 participants

## Key Eligibility Criteria

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### Inclusion (4)

- Participants must have a variant in ANKRD17 with a classification of VUS, likely pathogenic, or pathogenic
- Participants with a known diagnosis or CAGS have a disease-causing (likely pathogenic or pathogenic) variant in ANKRD17 evidenced by a pre-existing clinical genetic report.
- Participants with a suspected diagnosis of CAGS must have a variant of uncertain significance in ANKRD17 evidenced by a pre-existing clinical genetic report and clinical features of CAGS
- Participants with a VUS in ANKRD17 must have a variant of uncertain significance in ANKRD17

### Exclusion (1)

- No evidence of a disease-causing or potentially disease-causing variant ANRKD17 variant on a pre-existing clinical genetic report.

## Locations (1 total)

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Boston Children's Hospital, Boston, Massachusetts, United States