

Chopra-Amiel-Gordon Syndrome (CAGS) / ANKRD17 Study Opportunity

Natural history study of individuals with *ANKRD17* variants



WHO?

- Children and adults of all ages with a pathogenic variant in *ANKRD17* identified on genetic testing
- Children and adults of all ages with suspected CAGS based on a variant of uncertain significance (VUS) in *ANKRD17* and clinical features
- Children and adults of all ages with a VUS in *ANKRD17* whose diagnosis of CAGS is unclear



WHY?

- Better understand this rare genetic disorder with an observational study
- Investigate the spectrum of genetic variants in CAGS
- Characterize the clinical features of CAGS
- Generate patient-derived induced pluripotent stem cell (iPSC) lines for future therapeutic research



HOW?

- Clinician questionnaires
- Quality of life questionnaires
- Review of previous MRI scans
- Review of medical records
- Record genetic results
- In-person or remote visit(s) with the study team (~1-3 hours)
- Additional neurobehavioral assessments/questionnaires (~4-5 hours)
- Optional EEG(s)
- Optional blood/skin sample collection
- Optional photo collection

There is no benefit to participating in this research study. However, remote participation is permitted, and families will be compensated US \$50 for completion of the neurobehavioral assessment.

This research is being conducted by **Maya Chopra, MBBS, FRACP**

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