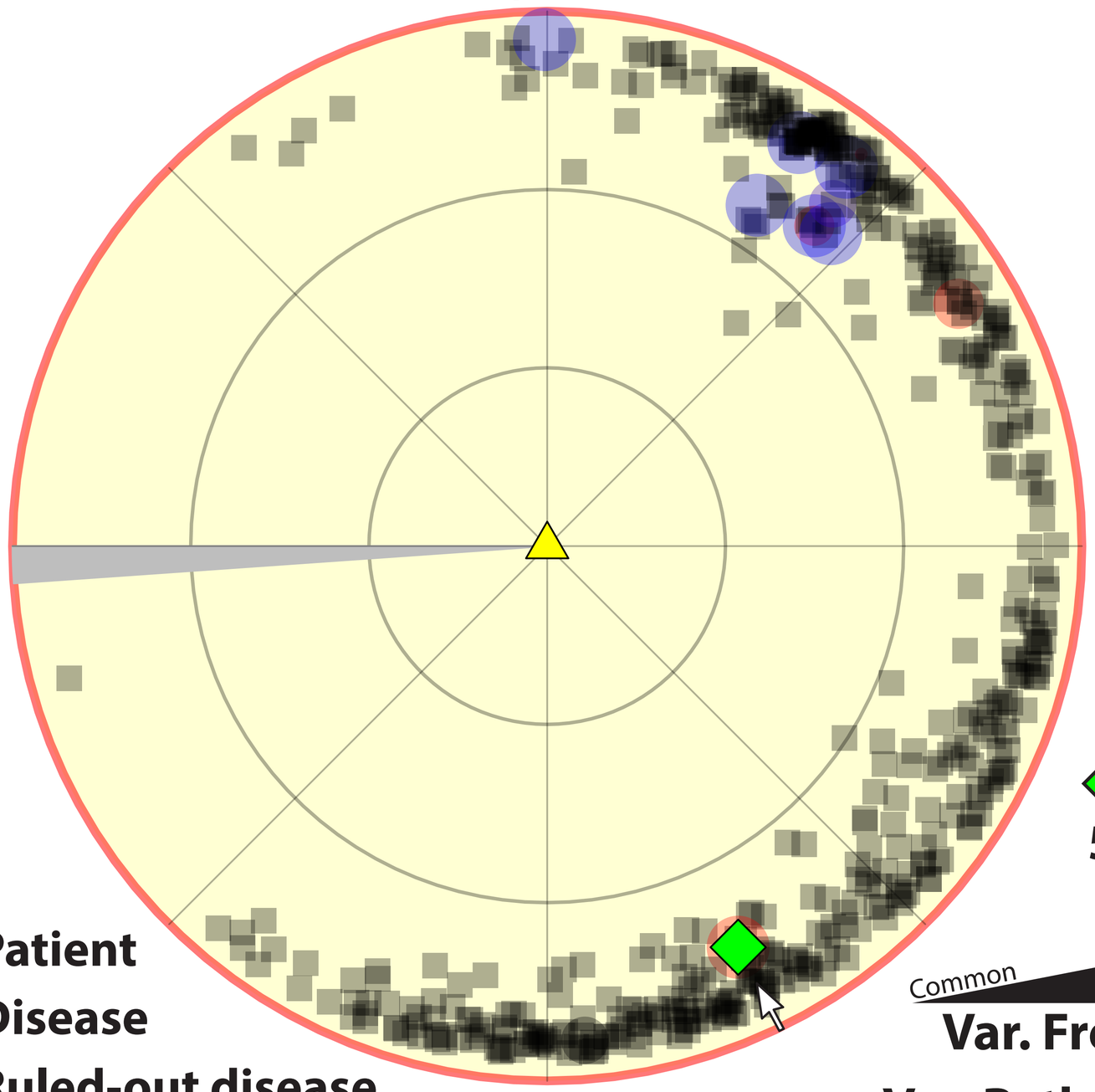
A



F

Hovered OMIM disease:
Epileptic encephalopathy, early infantile, 13 (614558)

Known inheritance:
Autosomal dominant

Relative similarity to query:
64.86%

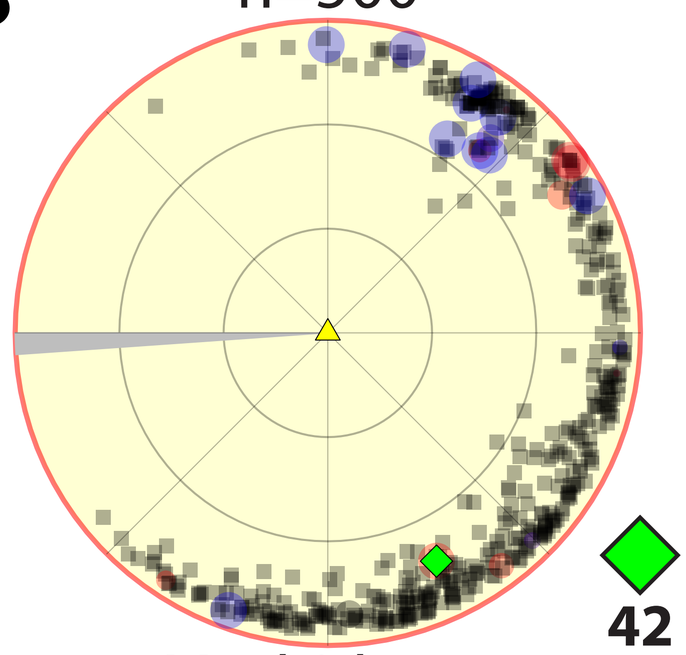
Variants uploaded in causal gene:
SCN8A | nonsynonymous | Thr->Ile | Chr12:52145307-52145307::C->T (ExAC freq=0.000e+00, pathogenicity=1)

Phenotypes shared with query:
Epileptic encephalopathy; Microcephaly; Intellectual disability; Seizures

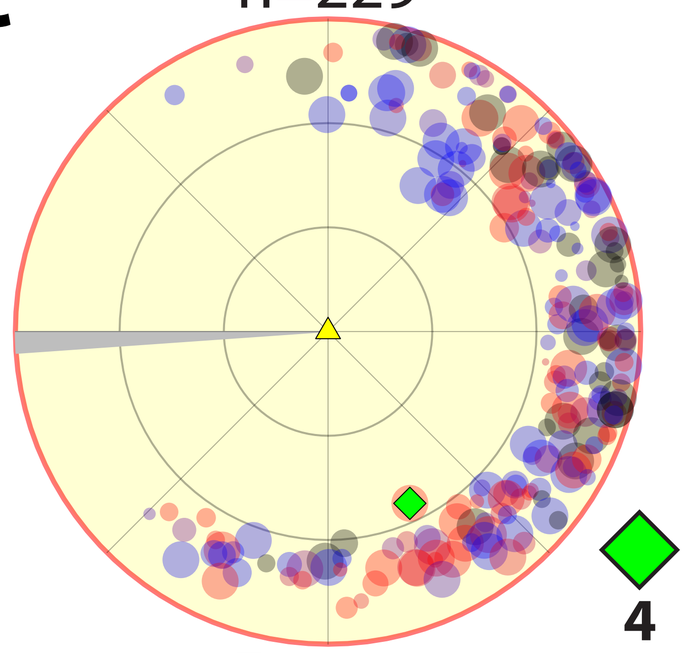
Query phenotypes not in disease:
Delayed CNS myelination; Gastroesophageal reflux; Pericardial effusion; Sinus bradycardia

Disease phenotypes not in query:
Autism; Cerebral atrophy; Developmental regression; Epileptic spasms...

B



C



G

Hovered OMIM disease:
Carpenter syndrome (201000)

Known inheritance:
Autosomal recessive

Relative similarity to query:
44.07%

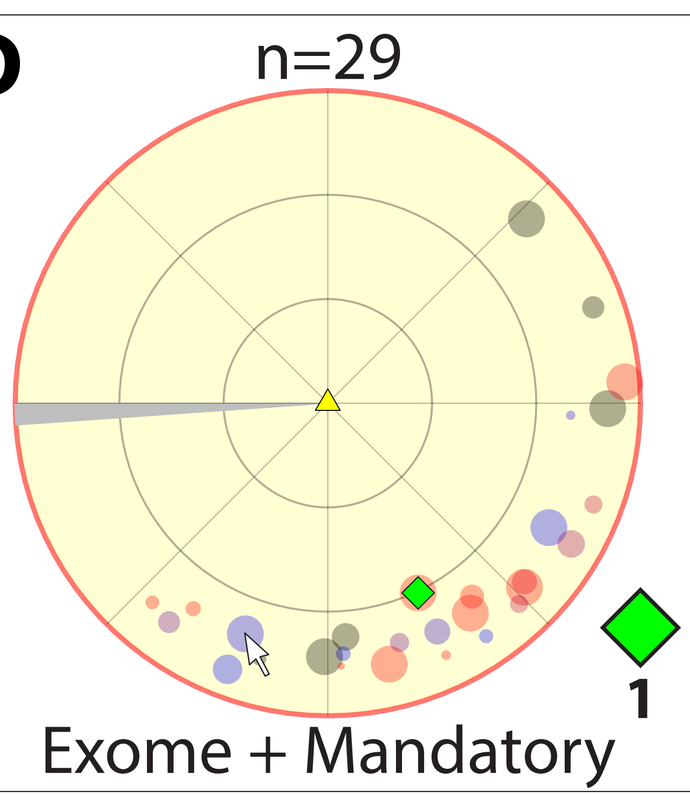
Variants uploaded in causal gene:
RAB23 | nonsynonymous | Ile->Leu | Chr6:57058685-57058685::T->G (ExAC freq=0.000e+00, pathogenicity=0.046209)

Phenotypes shared with query:
Aplasia/Hypoplasia of the cerebrum; Intellectual disability; Malformation of the heart and great vessels...

Query phenotypes not in disease:
Delayed CNS myelination; Epileptic encephalopathy; Pericardial effusion; Sinus bradycardia...

Disease phenotypes not in query:
Agenesis of permanent teeth; Aplasia/Hypoplasia of the middle phalanges...

D



E