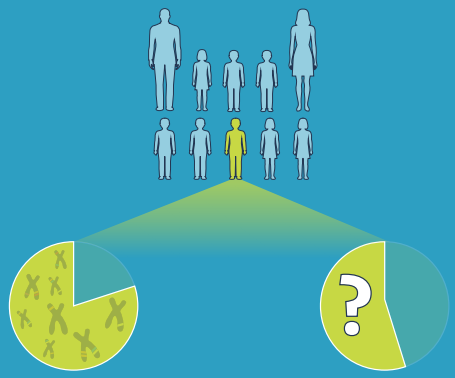
SOLVE MORE GENETIC DISEASES WITH LONG-READ SEQUENCING

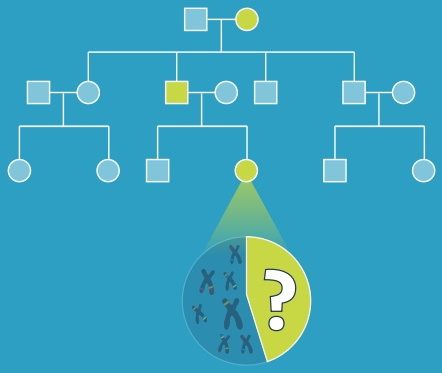
RARE DISEASES
affect **1** in **10** individuals



80%
are genetic in origin

>50%
of cases remain unsolved after short-read exome or WGS

MENDELIAN DISEASES
include over **8,500** known disorders



40%
have unknown genetic cause

DISEASES REMAIN UNSOLVED

STRUCTURAL VARIANTS ARE KNOWN TO CAUSE DISEASE
e.g. Schizophrenia, Carney Complex, Hereditary Breast & Ovarian Cancer

LONG READS
SENSITIVE TO ALL VARIANT TYPES

SHORT READS

SNVs (1 bp)	Indels (<50 bp)	Structural variants (≥50 bp)
5 Mb	3 Mb	10 Mb

variation between two human genomes by # of base pairs affected

ACCESS THE FULL SPECTRUM OF GENETIC VARIATION

INCREASE
SOLVE RATE

INCREASE
DISEASE GENE DISCOVERY

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