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

For Research Use Only. Not for use in diagnostic procedures.
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