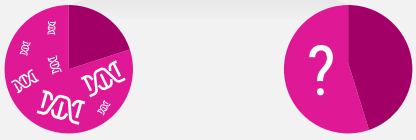
SHED LIGHT ON GENETIC DISEASES WITH LONG-READ SEQUENCING

RARE DISEASES

affect **1** in **10** individuals



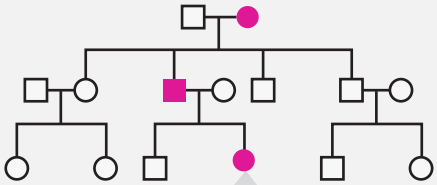
80%
are genetic in origin

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

DISEASES REMAIN UNEXPLAINED

MENDELIAN DISEASES

include over **8,500** known disorders



40%
have unknown genetic cause

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	INDELS	STRUCTURAL VARIANTS
(1 bp)	(<50 bp)	(≥50 bp)
5 Mb	3 Mb	10 Mb

Variation between two human genomes by number of base pairs affected

ACCESS THE FULL SPECTRUM OF GENETIC VARIATION

INCREASE
SOLVE RATE IN RARE DISEASE RESEARCH

INCREASE
DISEASE GENE DISCOVERY

PacBio