ACCESS THE FULL SPECTRUM OF GENETIC VARIATION

MENDELIAN DISEASES include over 8,500 known disorders

RARE DISEASES affect 1 in 10 individuals

80% are genetic in origin

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

40% have unknown genetic cause

>50% of cases remain unexplained

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

<table>
<thead>
<tr>
<th>SNP</th>
<th>INDEL</th>
<th>STRUCTURAL VARIANTS</th>
</tr>
</thead>
<tbody>
<tr>
<td>(1 bp)</td>
<td>(&lt;50 bp)</td>
<td>(≥50 bp)</td>
</tr>
<tr>
<td>5 Mb</td>
<td>3 Mb</td>
<td>10 Mb</td>
</tr>
</tbody>
</table>

Variation between two human genomes by number of base pairs affected

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INCREASE
- SOLVE RATE IN RARE DISEASE RESEARCH
- DISEASE GENE DISCOVERY