TYPES OF GENOMIC VARIATION

KNOWN DISEASES CAUSED BY STRUCTURAL VARIANTS



< 50 BASE PAIRS / VARIANT

SNV 1 BASE PAIR / VARIANT

> **INDELS** 1-49 BASE PAIRS / VARIANT

SCHIZOPHRENIA Deletion, duplication

CARNEY COMPLEX

Deletion

POOR DRUG METABOLISM

Duplication

HEREDITARY BREAST + OVARIAN CANCER

Deletion, insertion

NEUROFIBROMATOSIS

Insertion

CHRONIC MYELOID LEUKEMIA

Translocation

肃

DELETION

TANDEM DUPLICATION **STRUCTURAL**

VARIANTS ≥ 50 BASE PAIRS / VARIANT

> INTERSPERSED **DUPLICATION**

-≅

INVERSION

TRANSLOCATION

+ 1,000s MORE KNOWN...

...AND MANY TO BE **DISCOVERED**

STRUCTURAL VARIANTS **COMPRISE MORE THAN HALF** OF GENOMIC VARIATION

60%

> 20,000 STRUCTURAL **VARIANTS IN A HUMAN GENOME**

PacBi