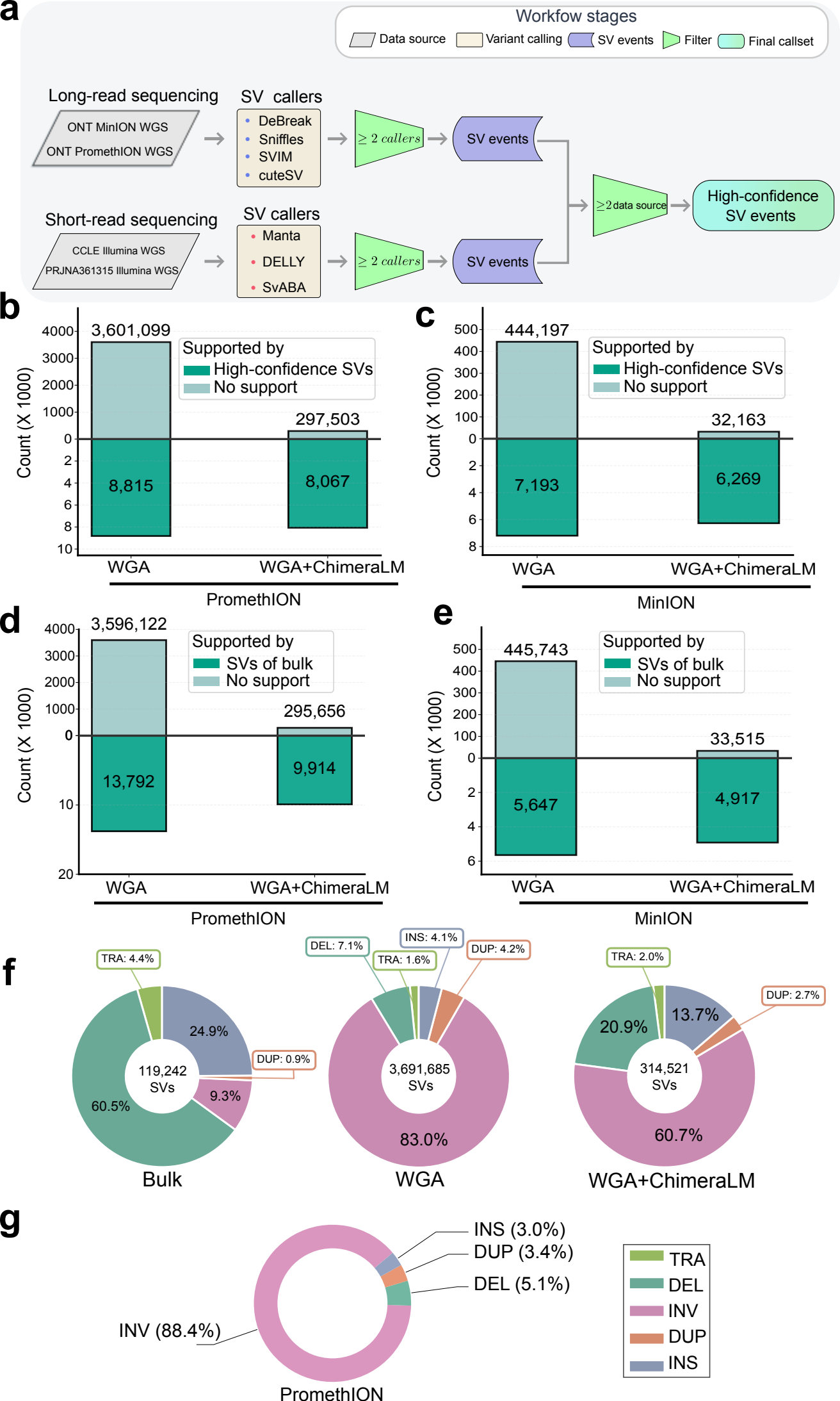
a
Workfow stages
Data source
Variant calling
SV events
Filter
Final callset
Long-read sequencing
ONT MinION WGS
ONT PromethION WGS
SV callers
DeBreak
Sniffles
SVIM
cuteSV
≥ 2 callers
SV events
Short-read sequencing
CCLE Illumina WGS
PRJNA361315 Illumina WGS
SV callers
Manta
DELLY
SvABA
≥ 2 callers
SV events
≥2 data source
High-confidence SV events
b
Count (X 1000)
3,601,099
8,815
297,503
8,067
Supported by
High-confidence SVs
No support
WGA
WGA+ChimeraLM
PromethION
c
444,197
7,193
32,163
6,269
Supported by
High-confidence SVs
No support
WGA
WGA+ChimeraLM
MinION
d
3,596,122
13,792
295,656
9,914
Supported by
SVs of bulk
No support
WGA
WGA+ChimeraLM
PromethION
e
445,743
5,647
33,515
4,917
Supported by
SVs of bulk
No support
WGA
WGA+ChimeraLM
MinION
f
TRA: 4.4%
24.9%
DUP: 0.9%
119,242 SVs
9.3%
60.5%
Bulk
DEL: 7.1%
INS: 4.1%
TRA: 1.6%
DUP: 4.2%
3,691,685 SVs
83.0%
WGA
TRA: 2.0%
DUP: 2.7%
13.7%
20.9%
314,521 SVs
60.7%
WGA+ChimeraLM
g
INS (3.0%)
DUP (3.4%)
DEL (5.1%)
INV (88.4%)
PromethION
TRA
DEL
INV
DUP
INS