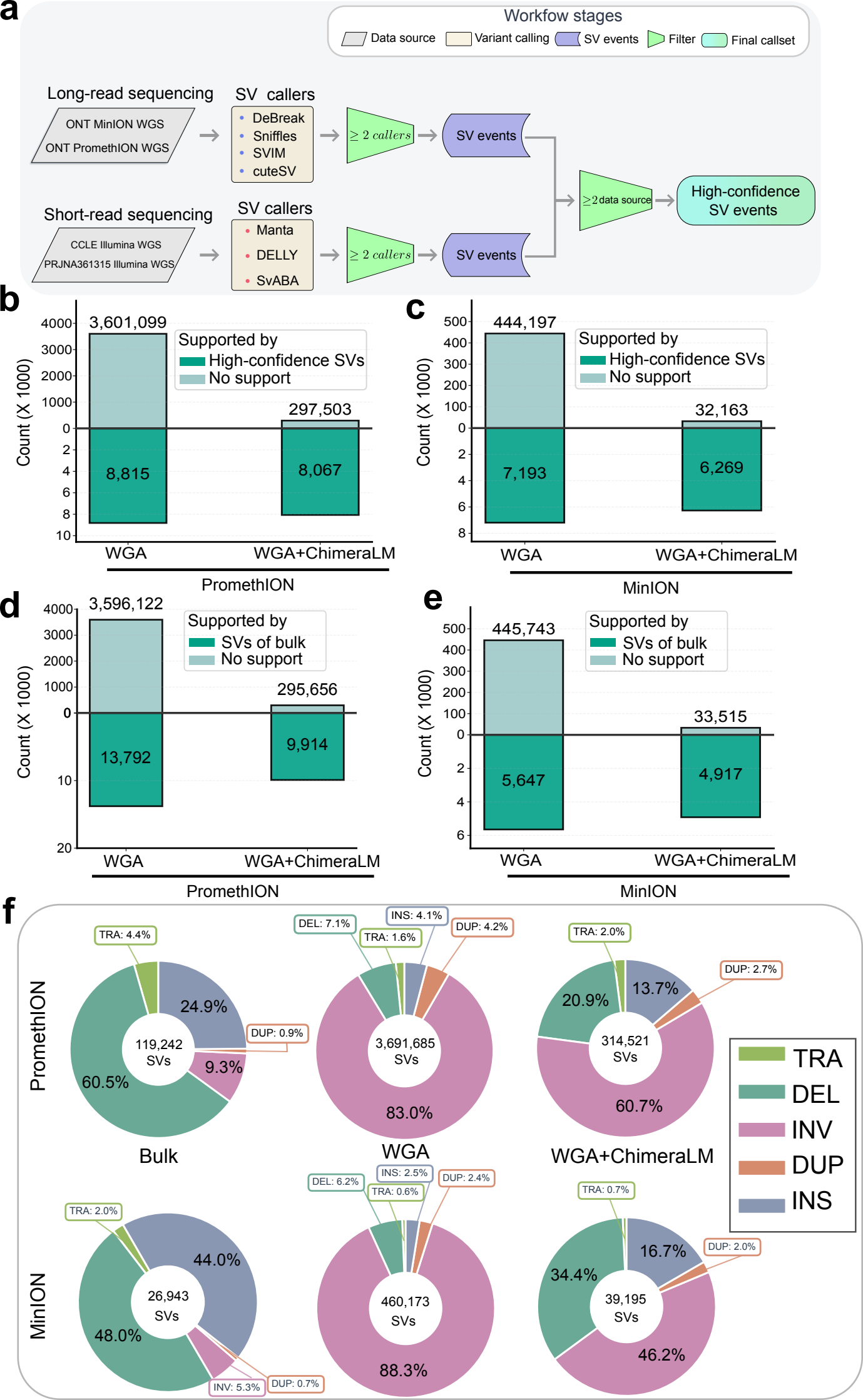
a
Workfow stages
Data source
Variant calling
SV events
Filter
Final callset
Long-read sequencing
ONT MinION WGS
ONT PromethION WGS
SV callers
DeBreak
Sniffles
SVIM
cuteSV
≥ 2 callers
SV events
Short-read sequencing
CCLE Illumina WGS
PRJNA361315 Illumina WGS
SV callers
Manta
DELLY
SvABA
≥ 2 callers
SV events
≥2 data source
High-confidence SV events
b
Count (X 1000)
3,601,099
Supported by
High-confidence SVs
No support
297,503
8,815
8,067
WGA
WGA+ChimeraLM
PromethION
c
444,197
Supported by
High-confidence SVs
No support
32,163
7,193
6,269
WGA
WGA+ChimeraLM
MinION
d
3,596,122
Supported by
SVs of bulk
No support
295,656
13,792
9,914
WGA
WGA+ChimeraLM
PromethION
e
445,743
Supported by
SVs of bulk
No support
33,515
5,647
4,917
WGA
WGA+ChimeraLM
MinION
f
PromethION
MinION
Bulk
WGA
WGA+ChimeraLM
TRA
DEL
INV
DUP
INS
119,242 SVs
TRA: 4.4%
24.9%
DUP: 0.9%
9.3%
60.5%
3,691,685 SVs
DEL: 7.1%
INS: 4.1%
TRA: 1.6%
DUP: 4.2%
83.0%
314,521 SVs
TRA: 2.0%
DUP: 2.7%
20.9%
13.7%
60.7%
26,943 SVs
TRA: 2.0%
44.0%
48.0%
INV: 5.3%
DUP: 0.7%
460,173 SVs
DEL: 6.2%
INS: 2.5%
TRA: 0.6%
DUP: 2.4%
88.3%
39,195 SVs
TRA: 0.7%
16.7%
DUP: 2.0%
34.4%
46.2%