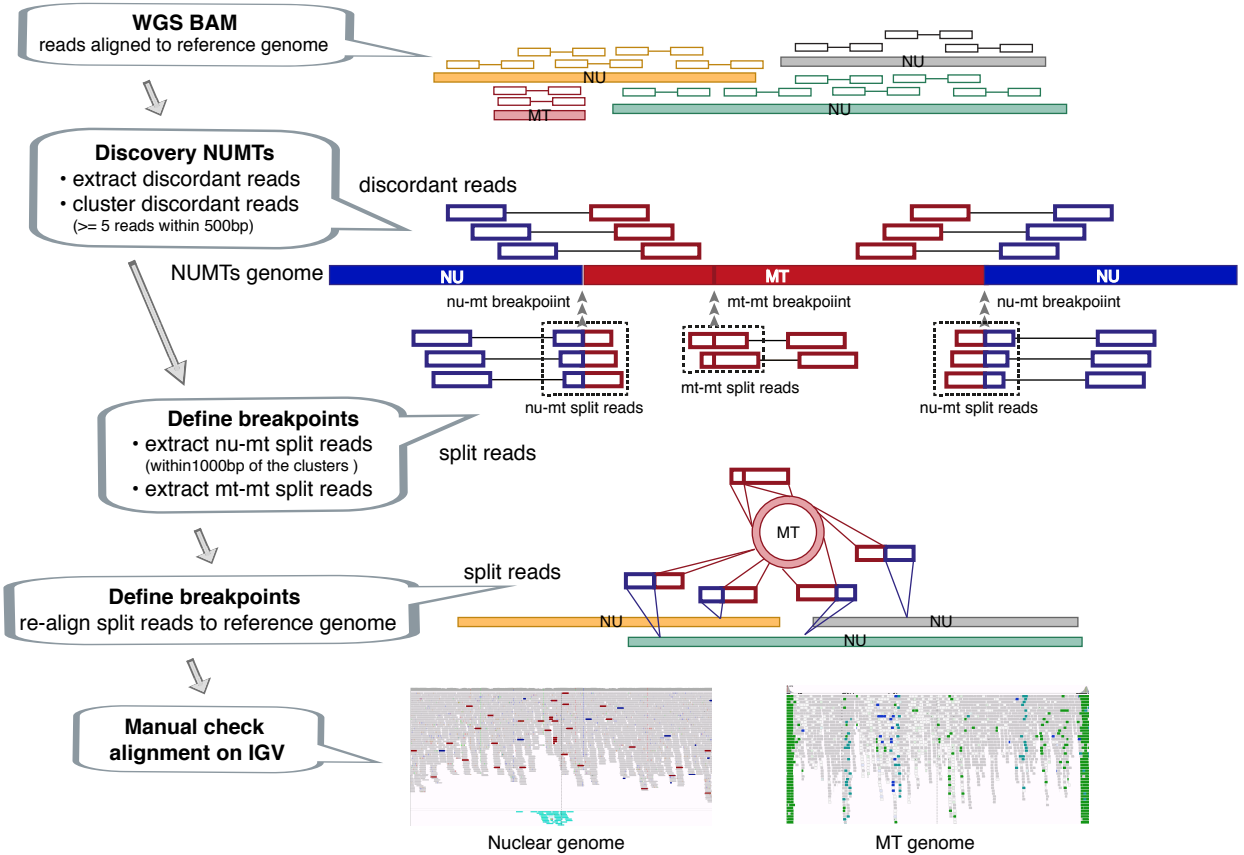
WGS BAM
reads aligned to reference genome
NU
NU
MT
NU
Discovery NUMTs
• extract discordant reads
• cluster discordant reads
(>= 5 reads within 500bp)
discordant reads
NUMTs genome
NU
MT
NU
nu-mt breakpoiint
mt-mt breakpoiint
nu-mt breakpoiint
nu-mt split reads
mt-mt split reads
nu-mt split reads
Define breakpoints
• extract nu-mt split reads
(within1000bp of the clusters)
• extract mt-mt split reads
split reads
Define breakpoints
re-align split reads to reference genome
split reads
MT
NU
NU
NU
Manual check
alignment on IGV
Nuclear genome
MT genome