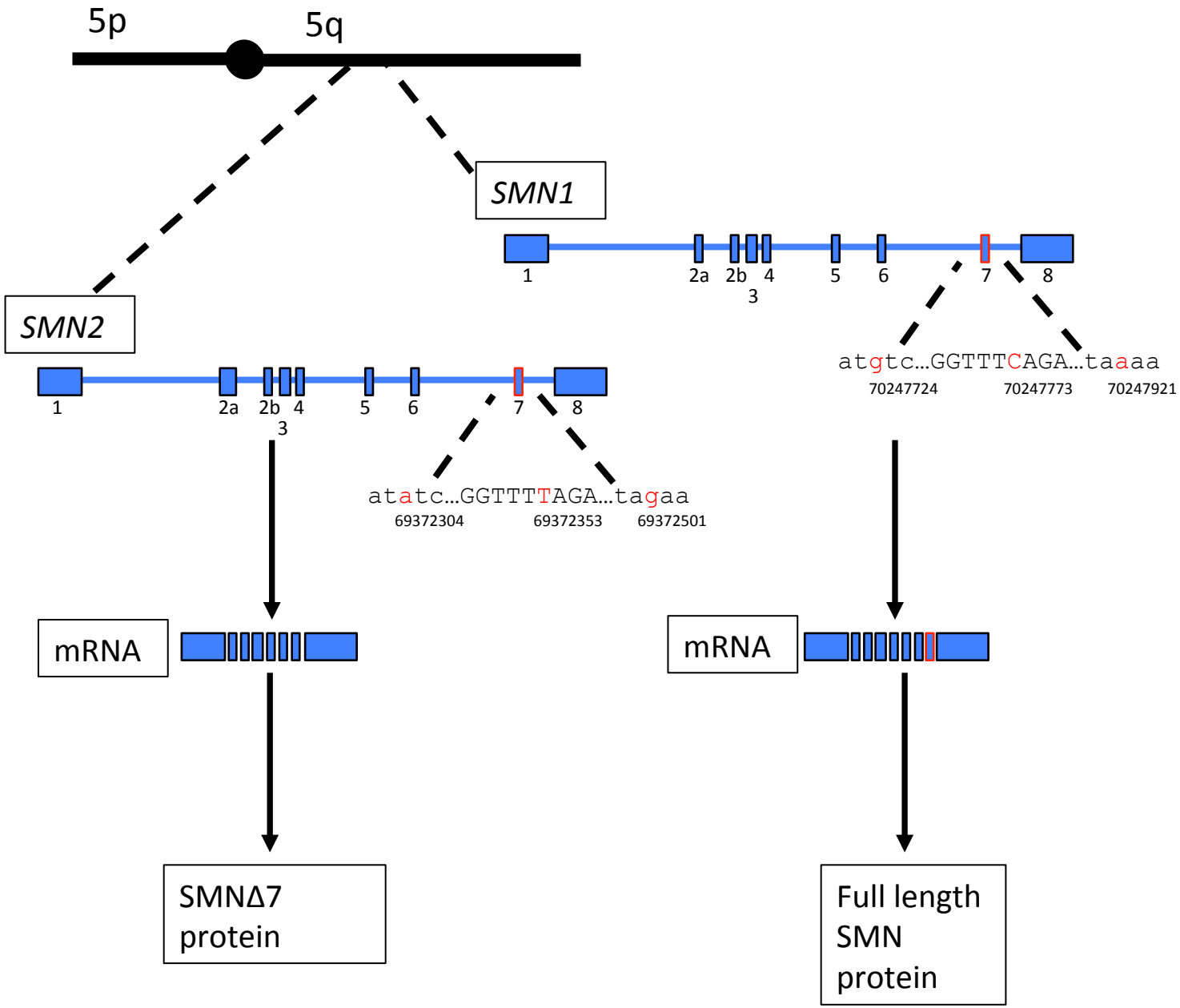
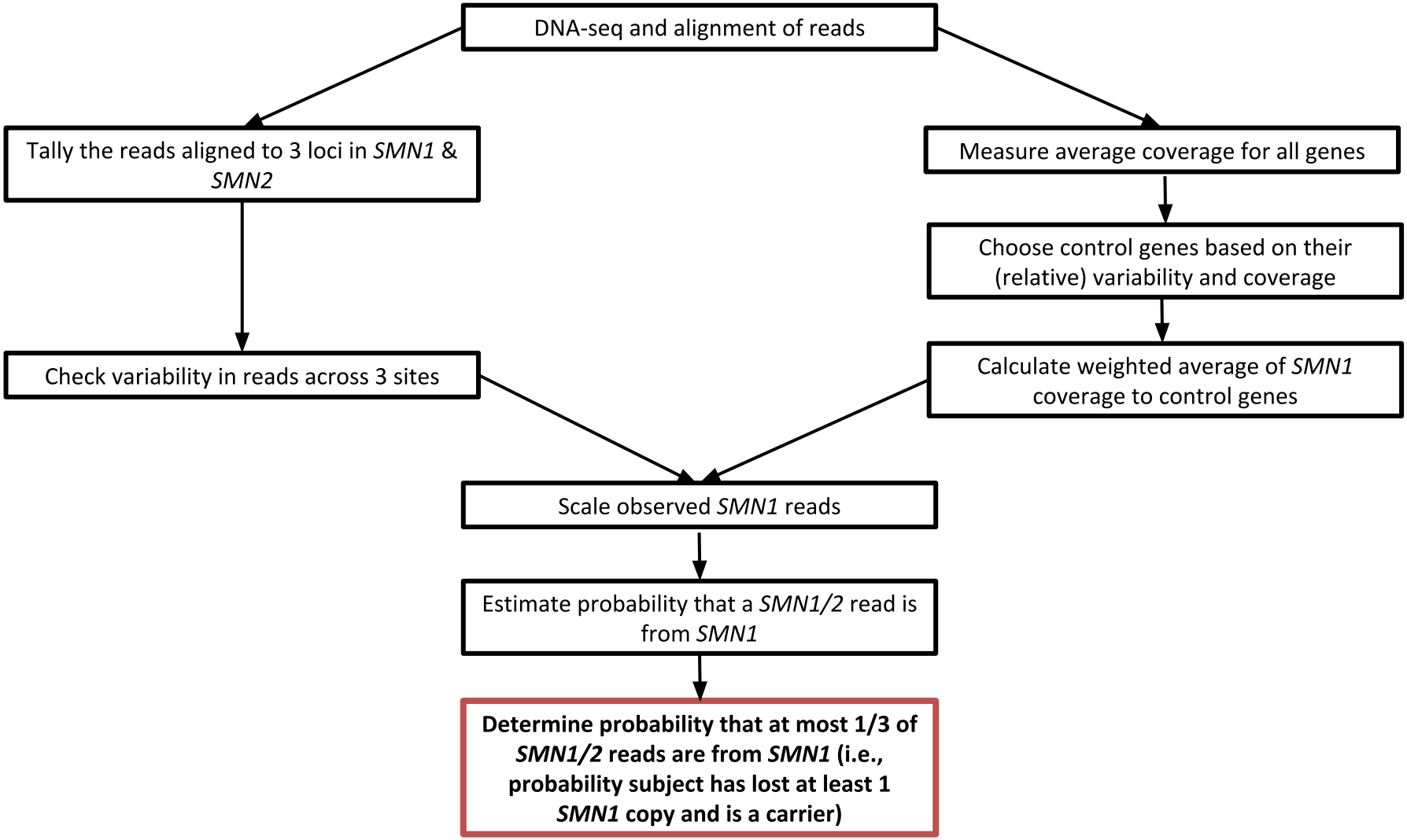

5p
5q
SMN1
SMN2
1
2a
2b
3
4
5
6
7
8
atgtc…GGTTTCAGA…taaaa
70247724
70247773
70247921
atatc…GGTTTTAGA…tagaa
69372304
69372353
69372501
mRNA
mRNA
SMNΔ7 protein
Full length SMN protein


DNA-seq and alignment of reads
Tally the reads aligned to 3 loci in *SMN1* & *SMN2*
Measure average coverage for all genes
Choose control genes based on their (relative) variability and coverage
Check variability in reads across 3 sites
Calculate weighted average of *SMN1* coverage to control genes
Scale observed *SMN1* reads
Estimate probability that a *SMN1/2* read is from *SMN1*
**Determine probability that at most 1/3 of *SMN1/2* reads are from *SMN1* (i.e., probability subject has lost at least 1 *SMN1* copy and is a carrier)**