

FIG. 1A

Mutations causing SMA

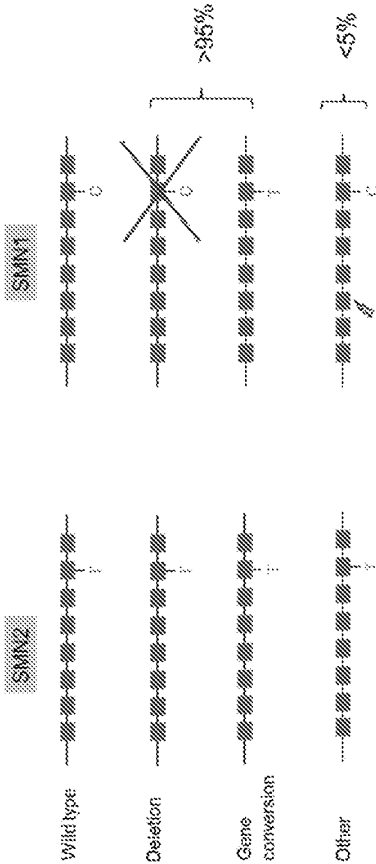


FIG. 1B

Common CNVs affecting SMN genes

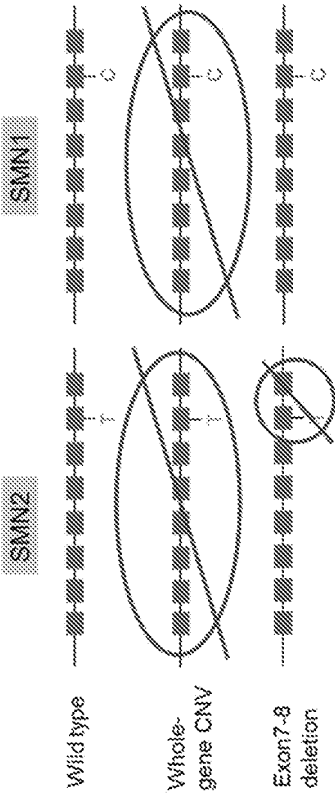


FIG. 1C

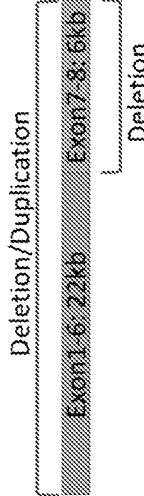


FIG. 1D

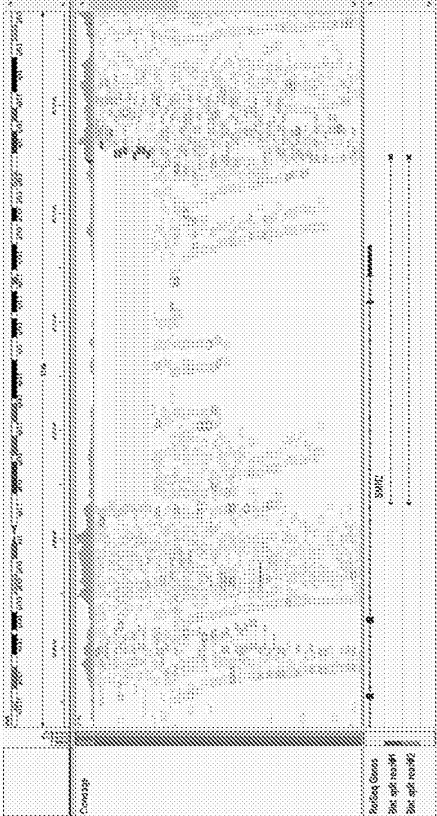
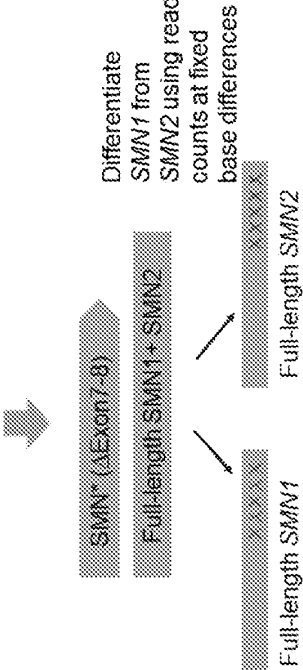
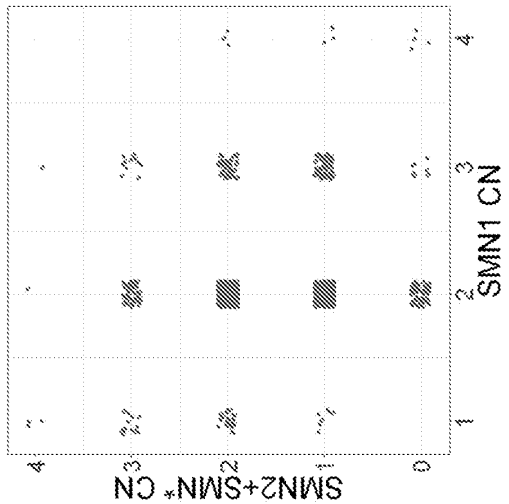
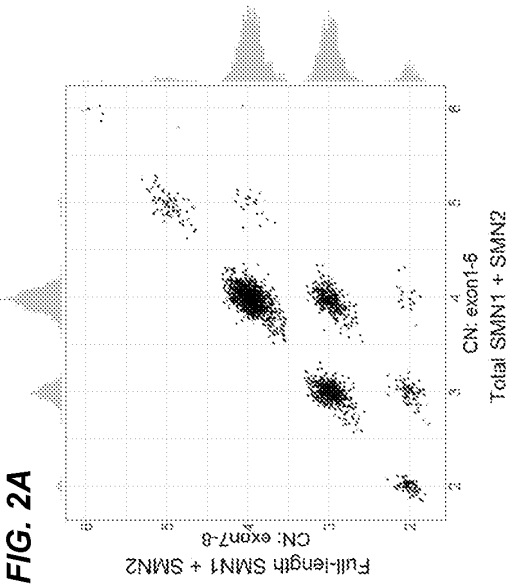
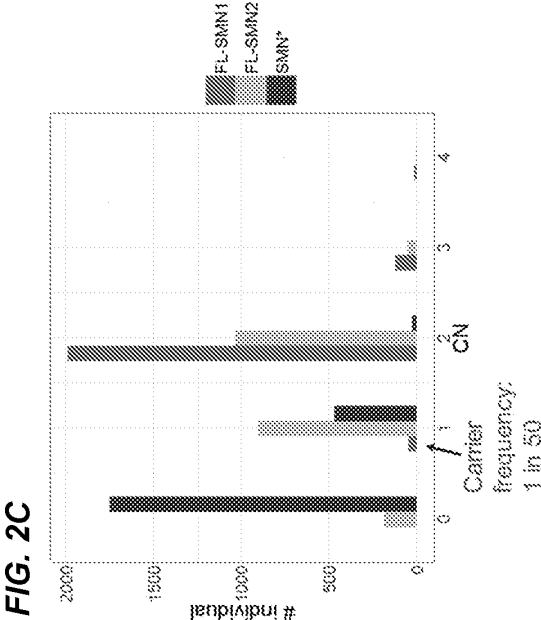


FIG. 1E

Call SMN1 + SMN2 copy number in two regions, Exon1-6 and Exon7-8, by summing up reads in SMN1 and SMN2 (no need for realignment to modified reference)





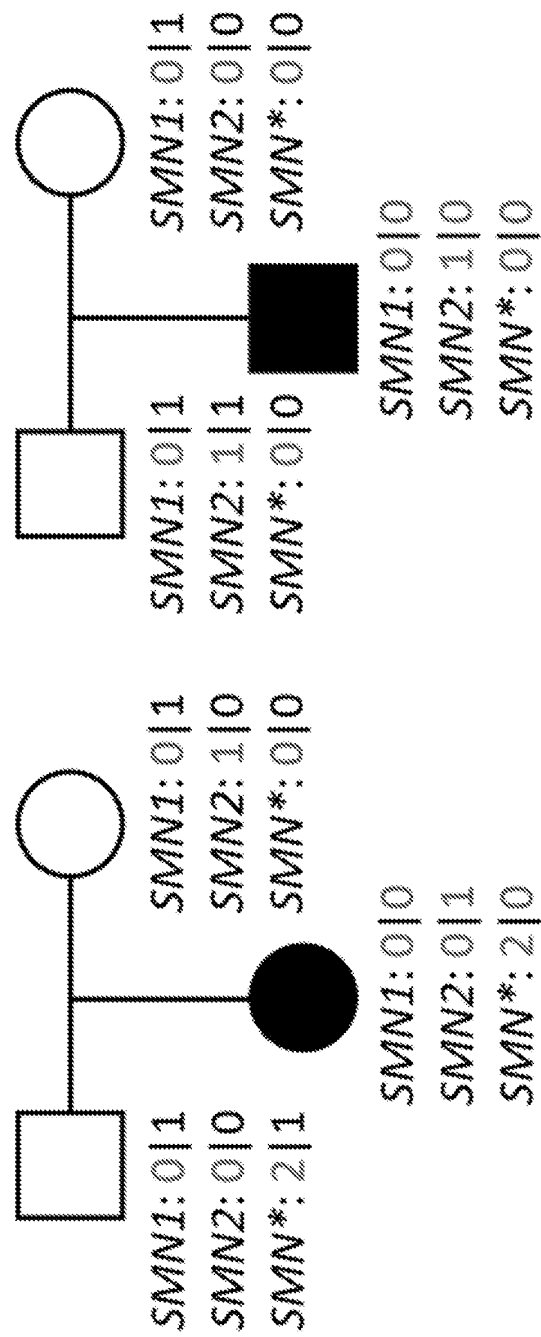
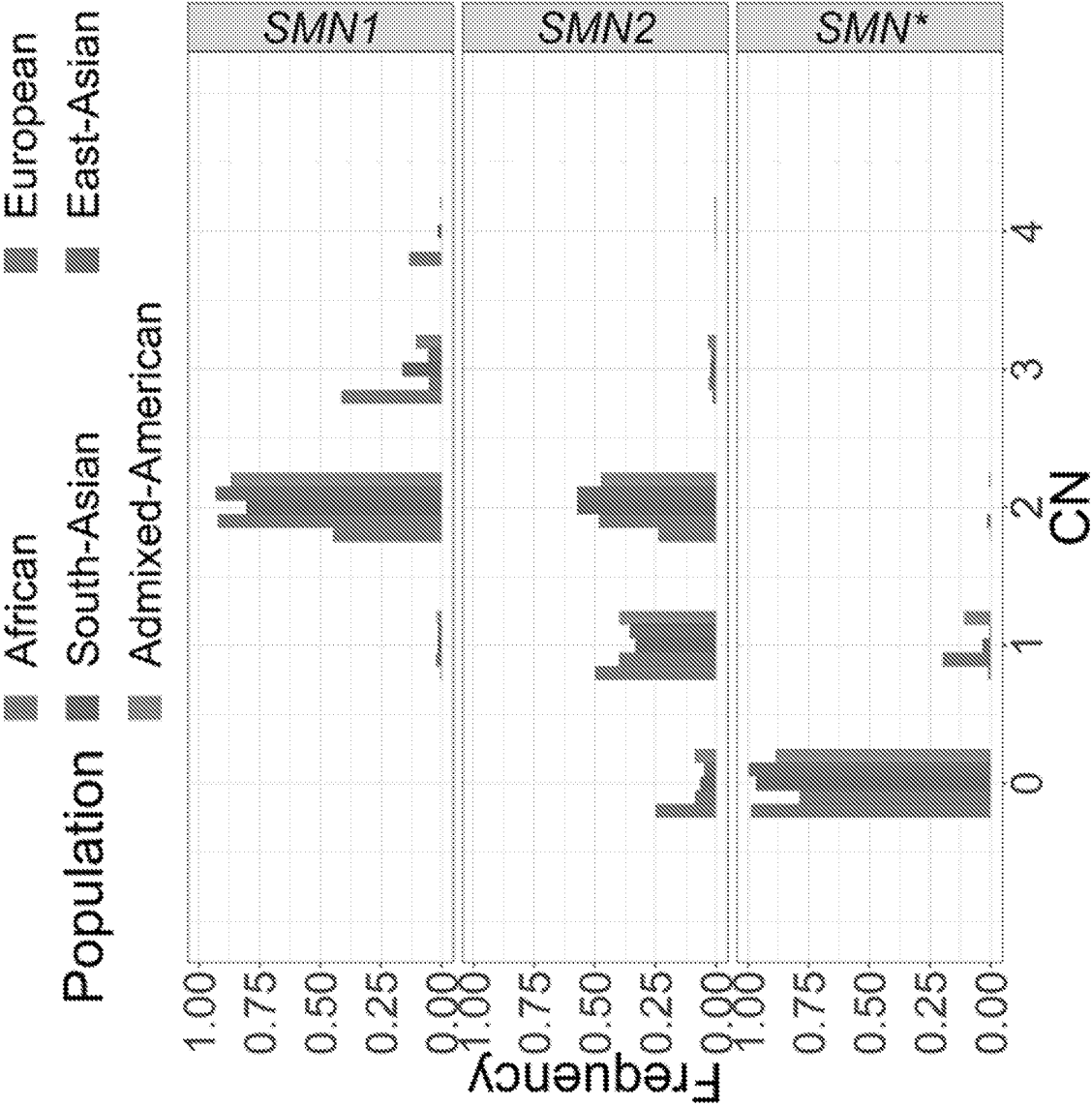


FIG. 3

FIG. 4



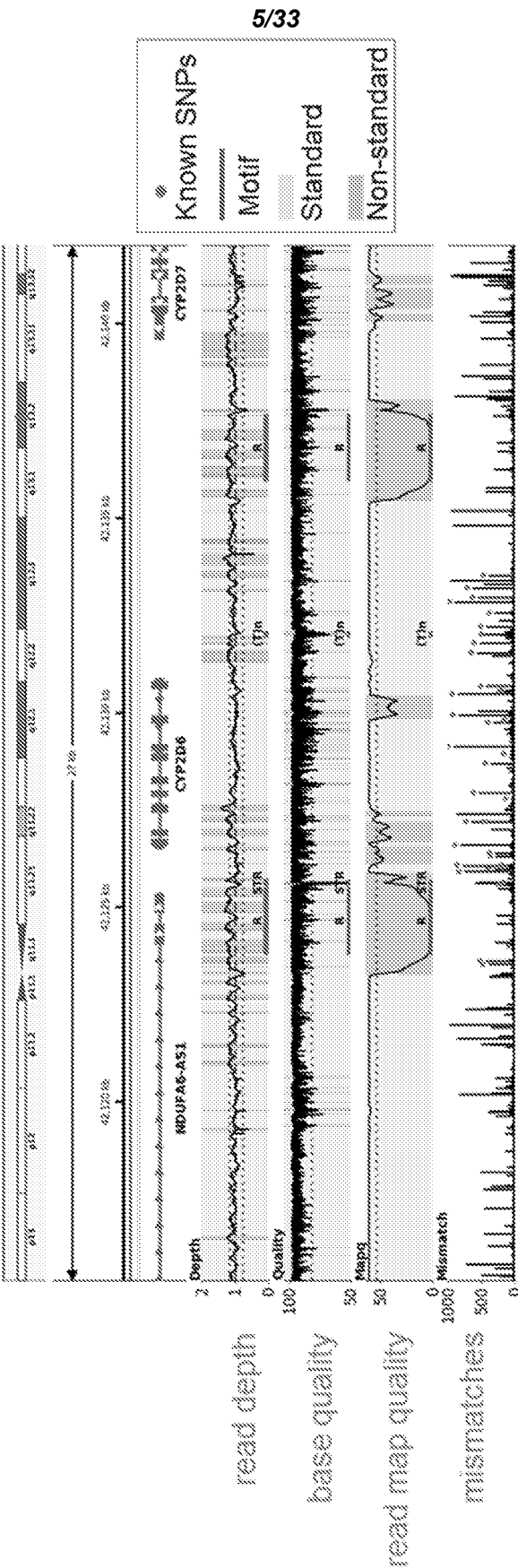


FIG. 5

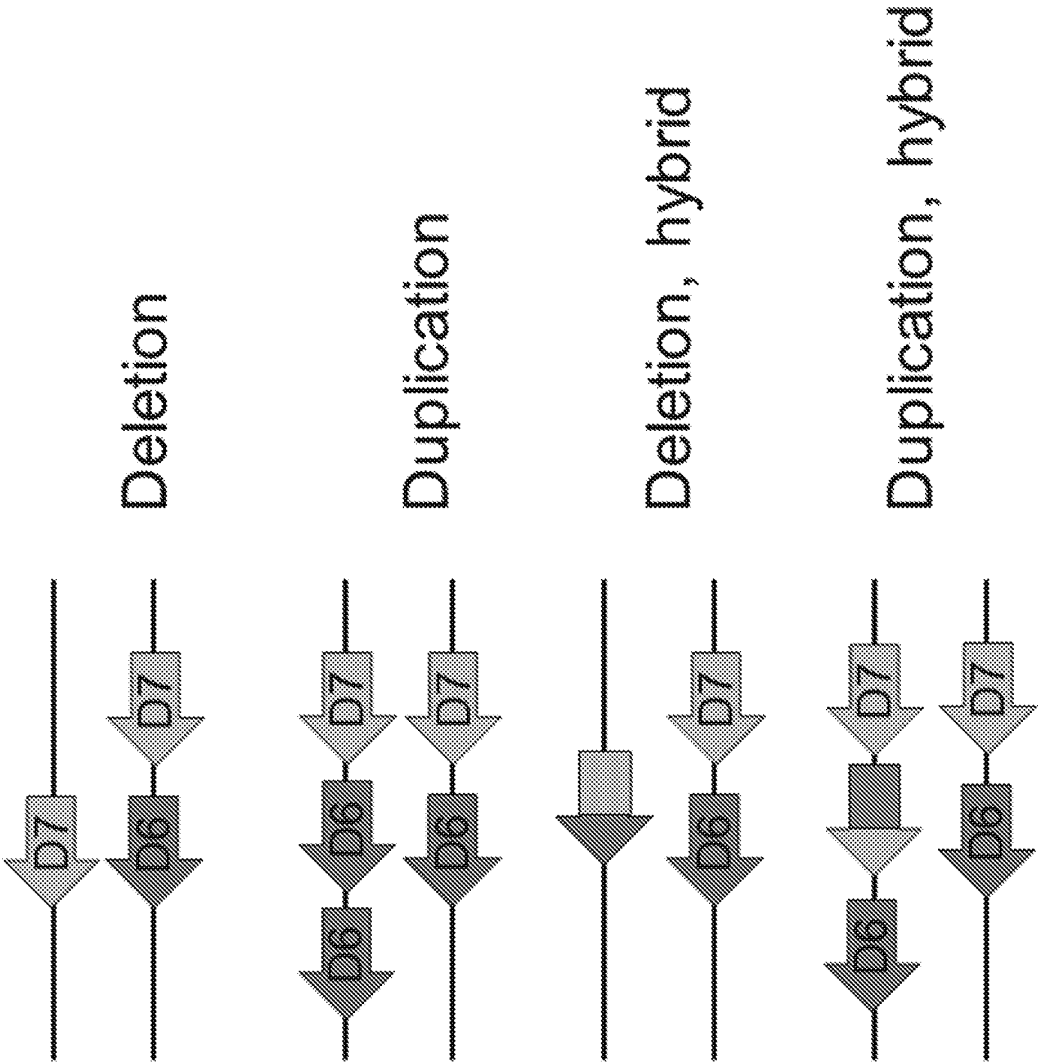


FIG. 6

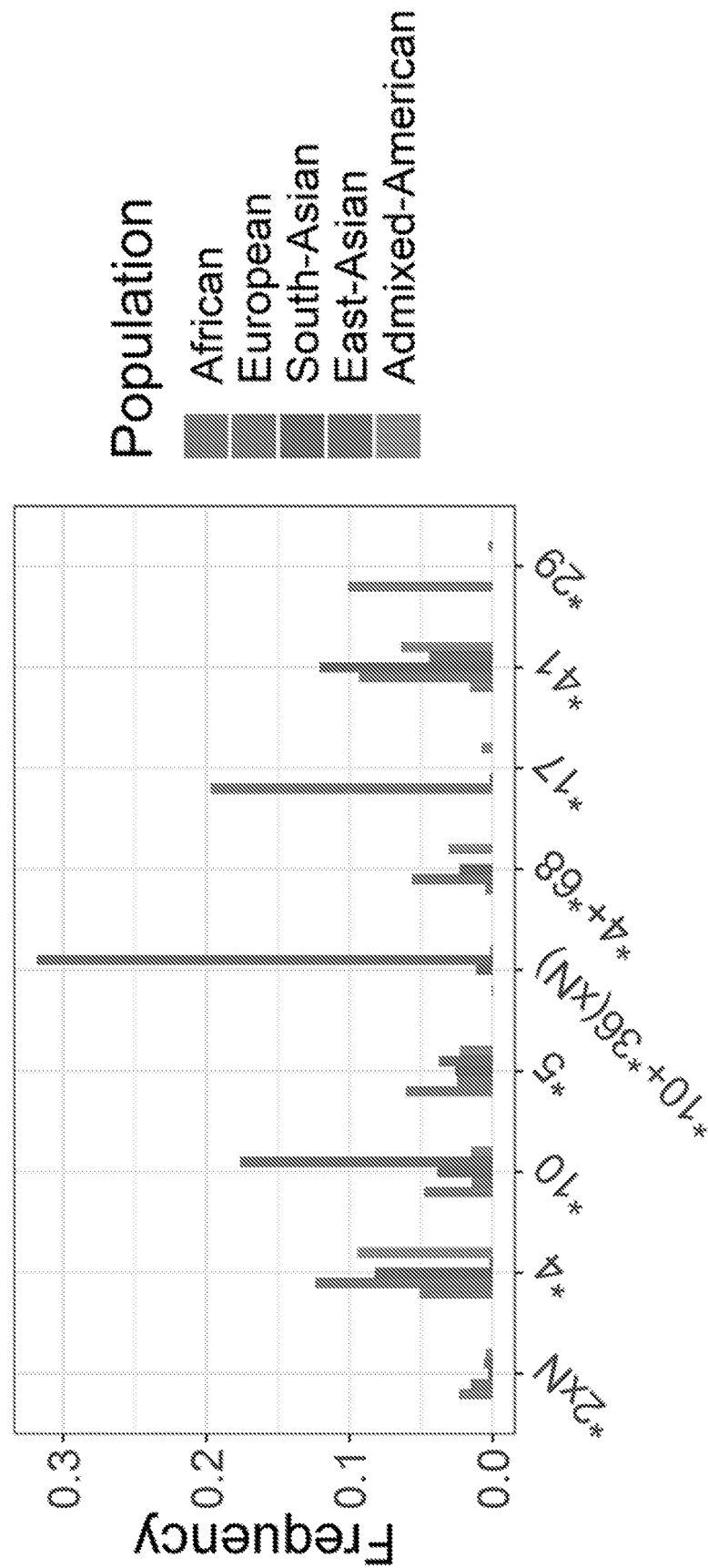
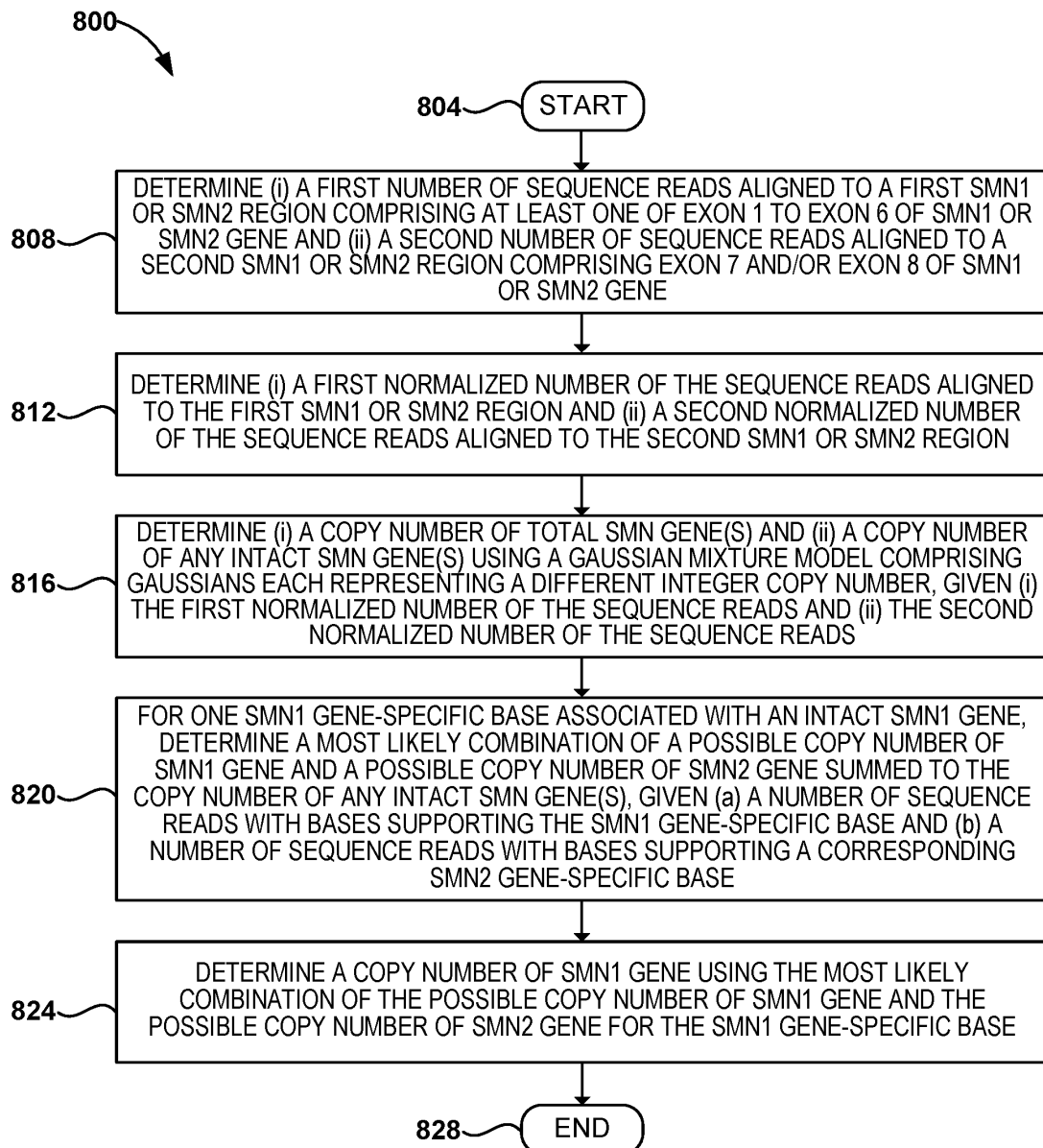
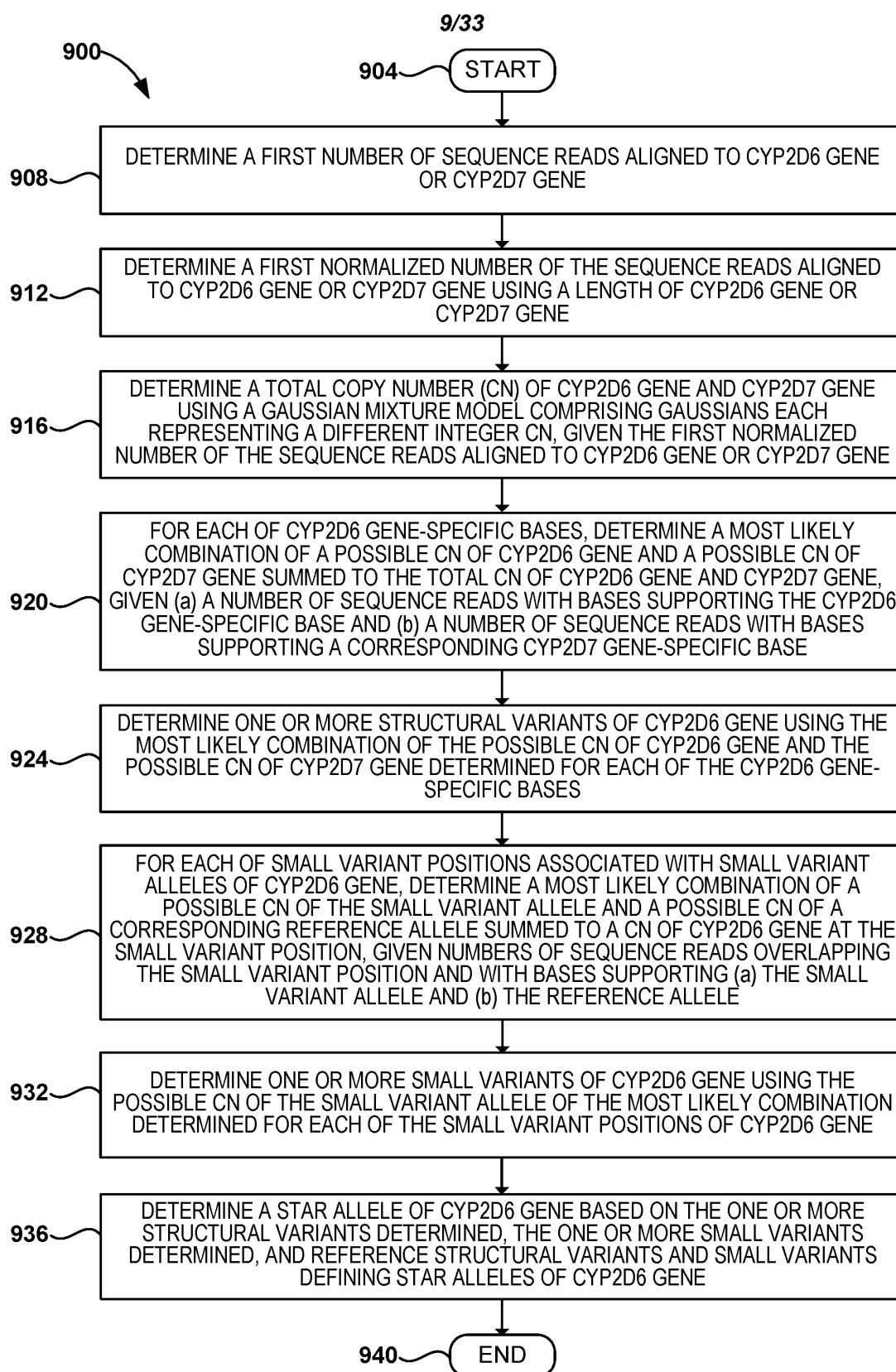


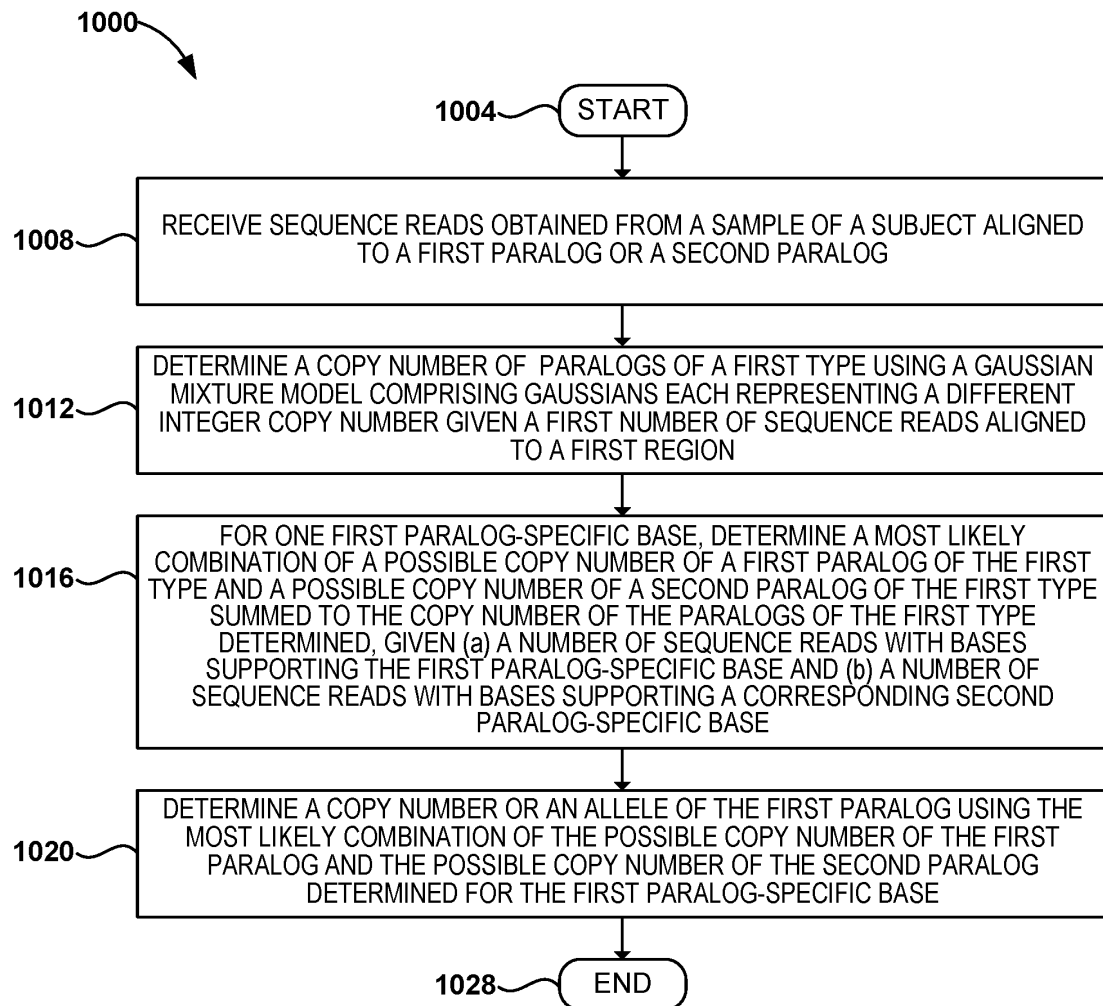
FIG. 7

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**FIG. 8**

**FIG. 9**

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**FIG. 10**

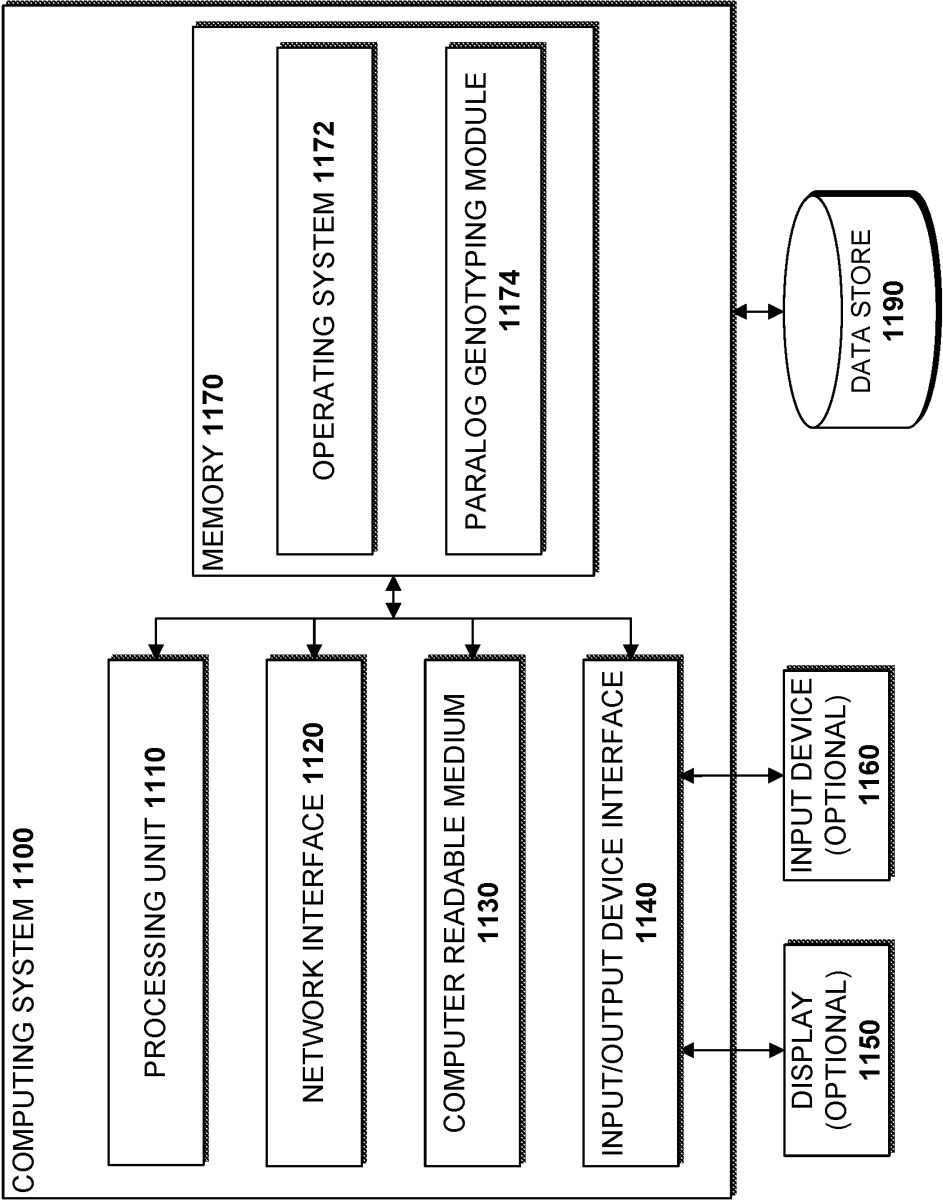


FIG. 11

FIG. 12A

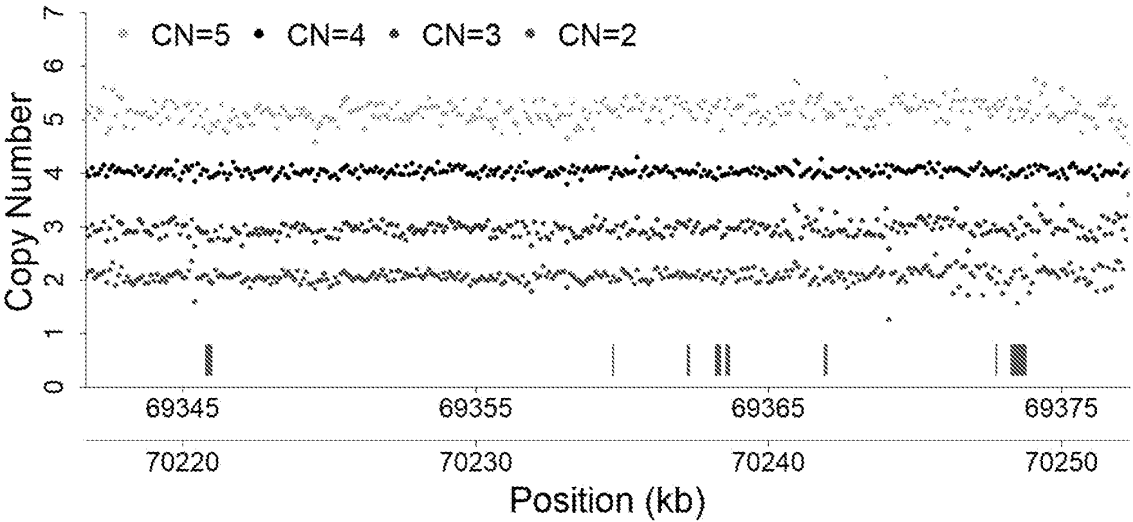
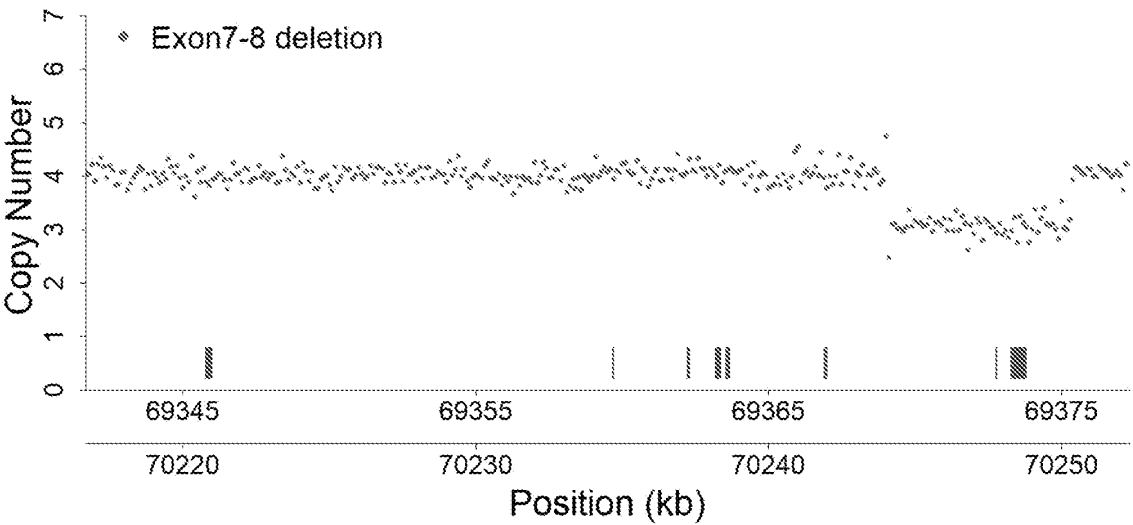


FIG. 12B



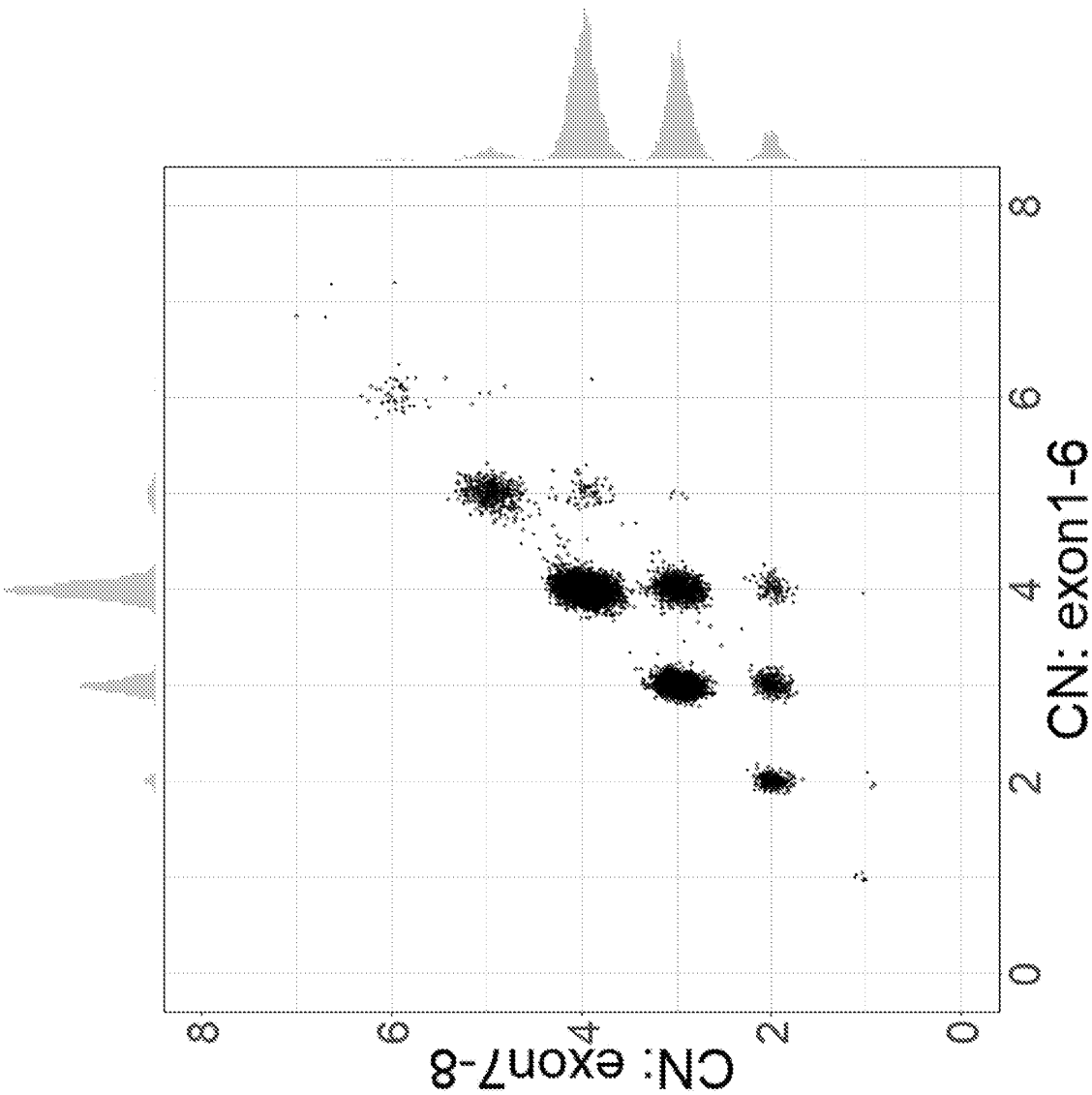


FIG. 13

FIG. 14A

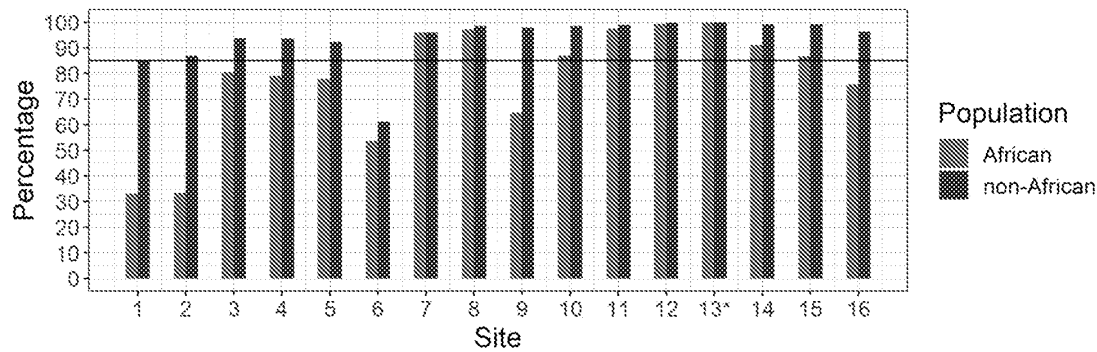


FIG. 14B

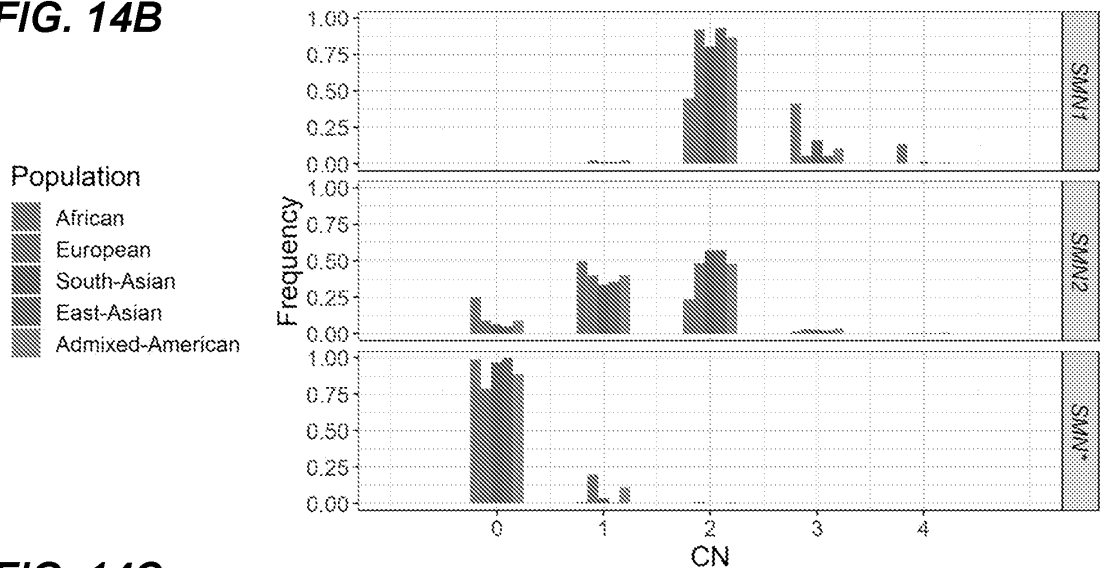


FIG. 14C

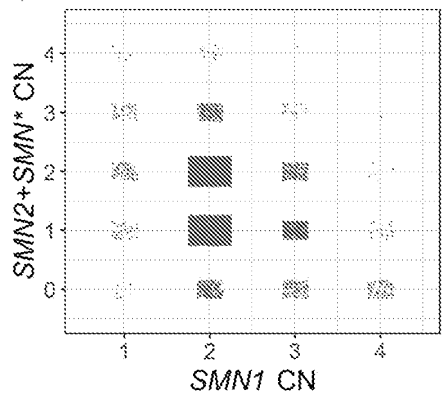


FIG. 14D

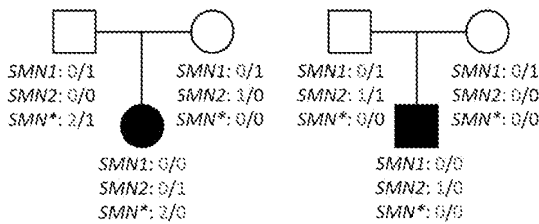
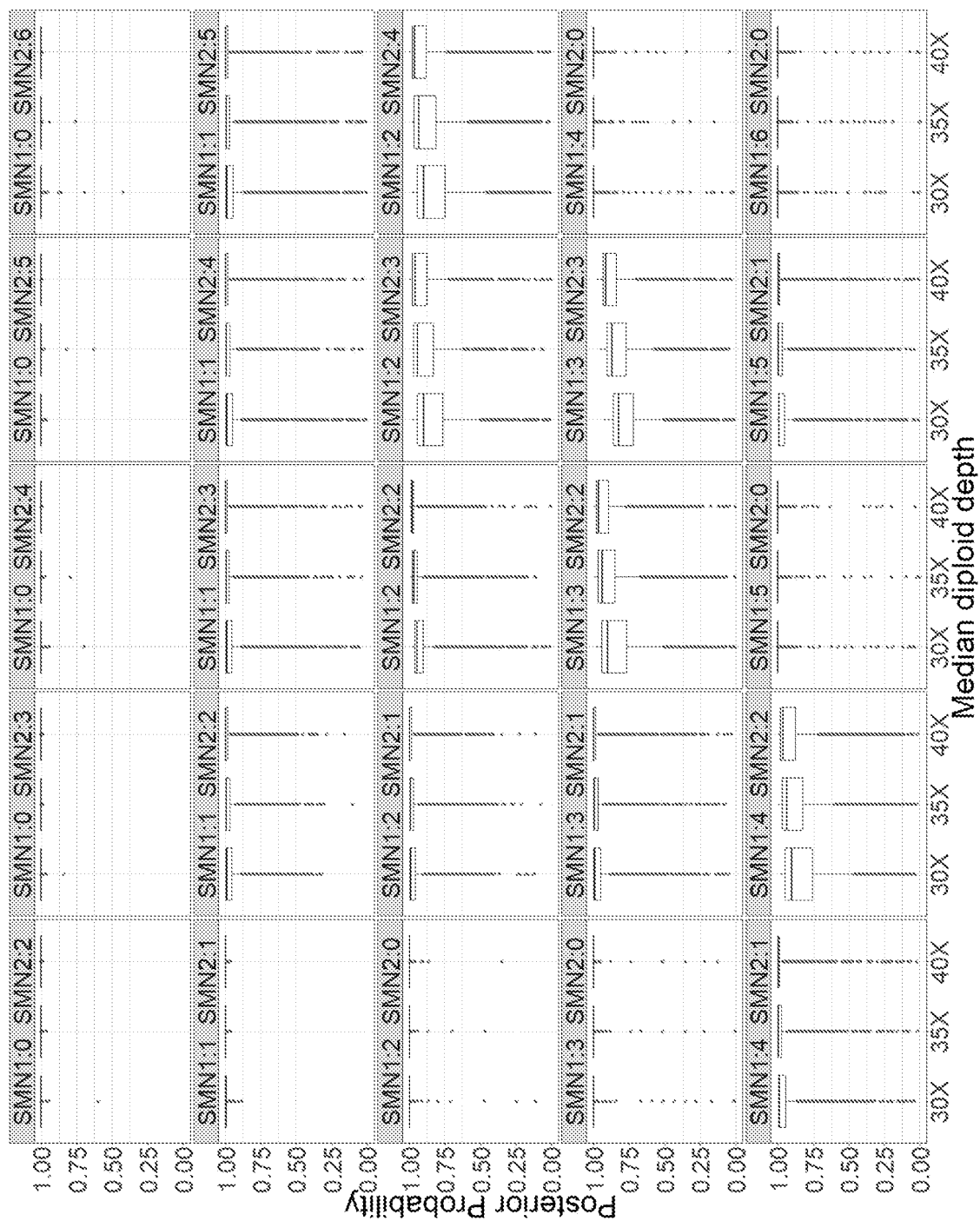


FIG. 15



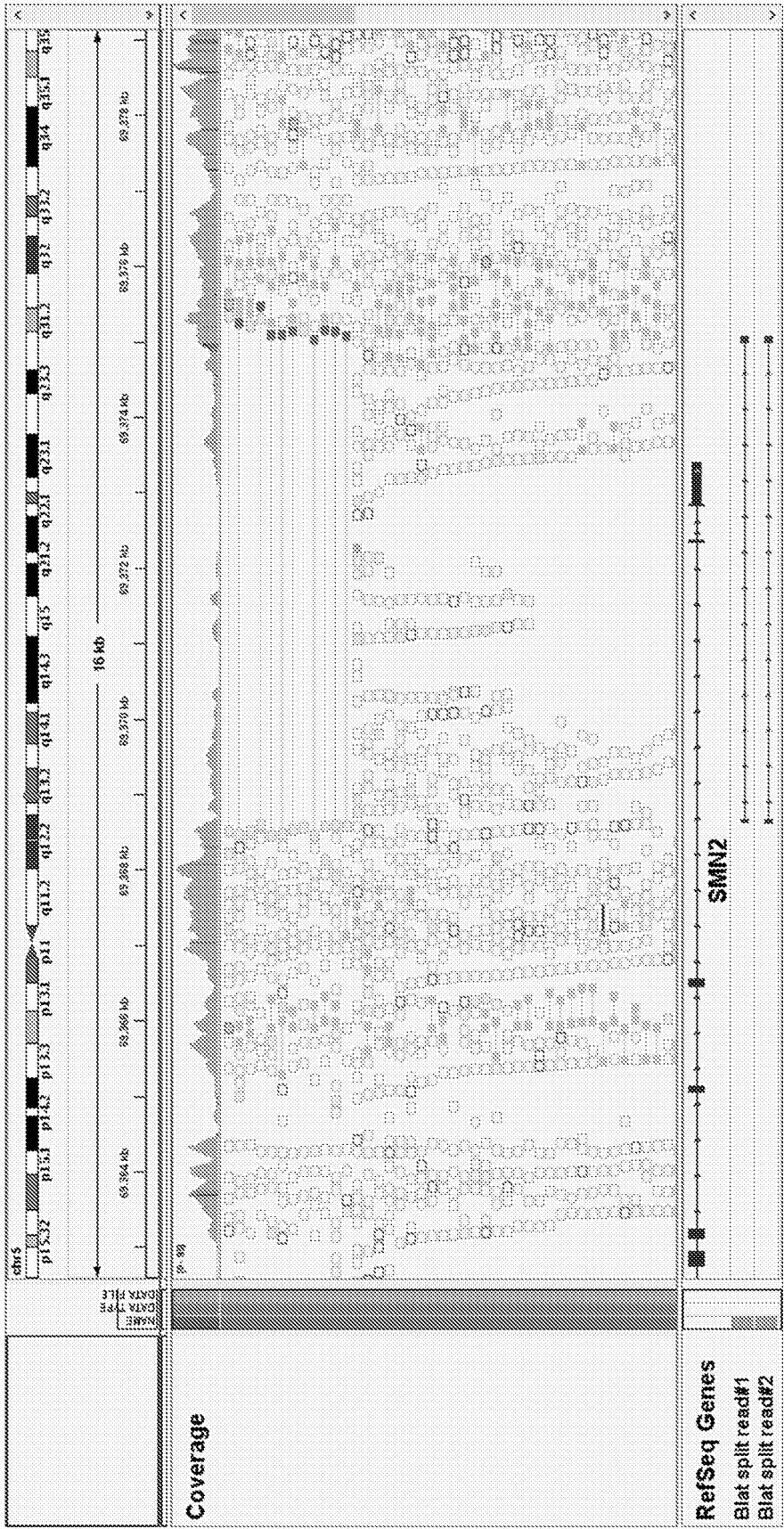


FIG. 16

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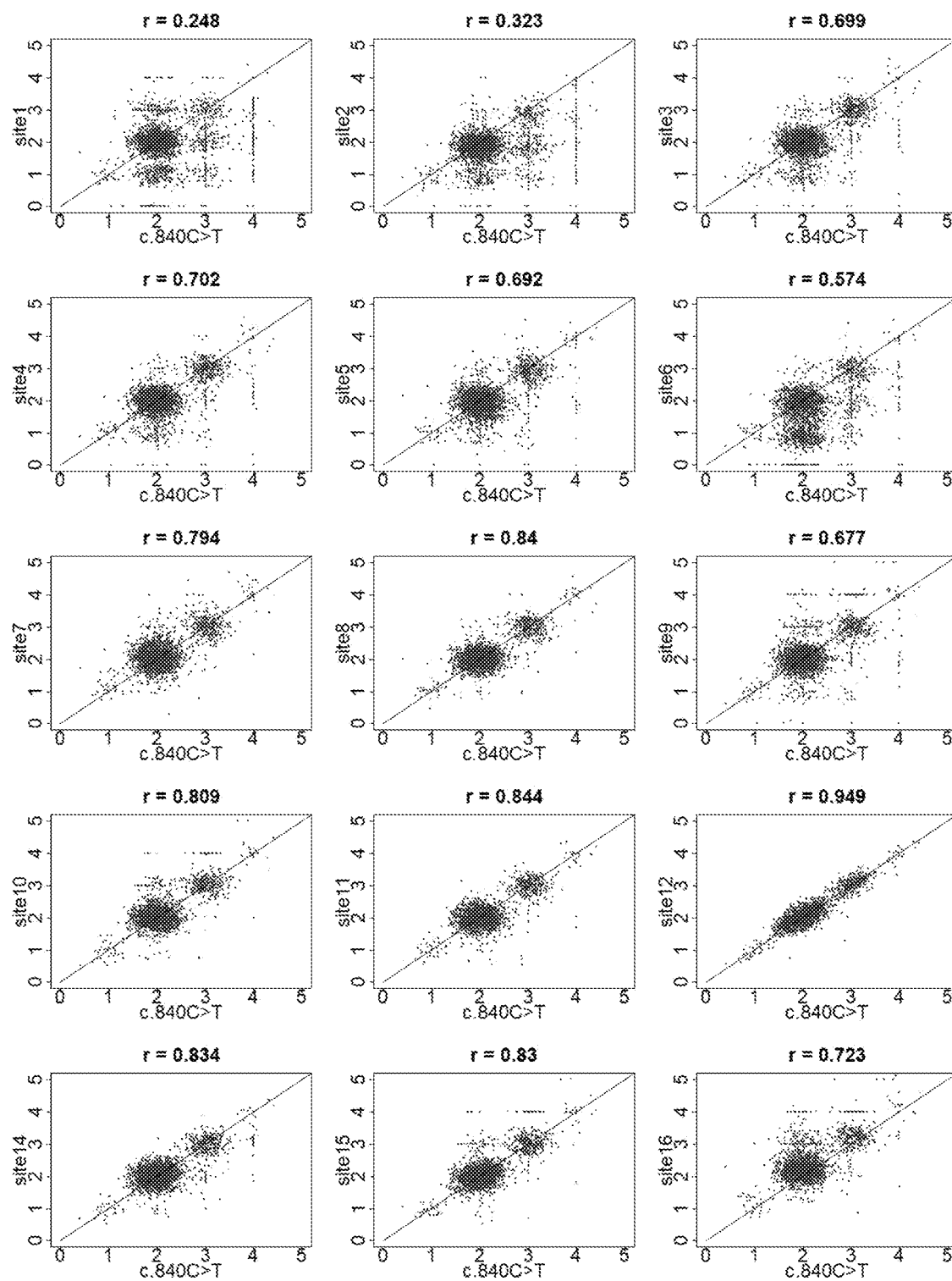
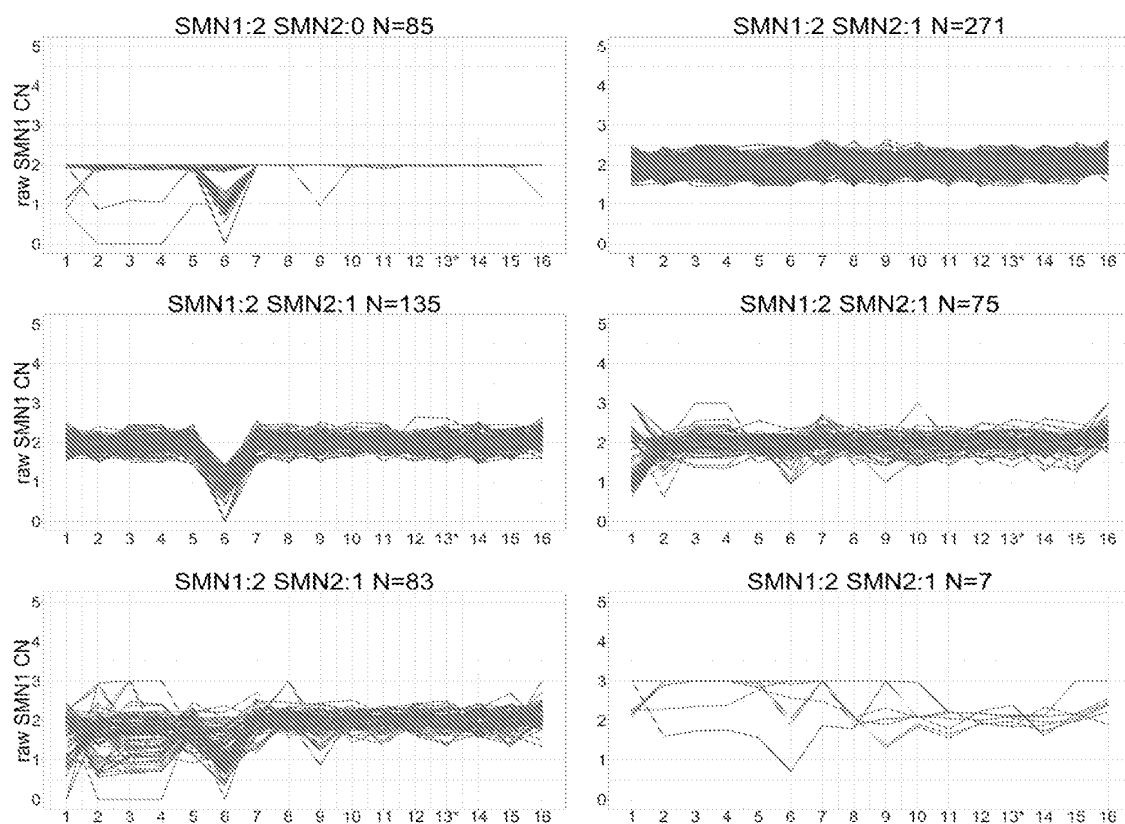
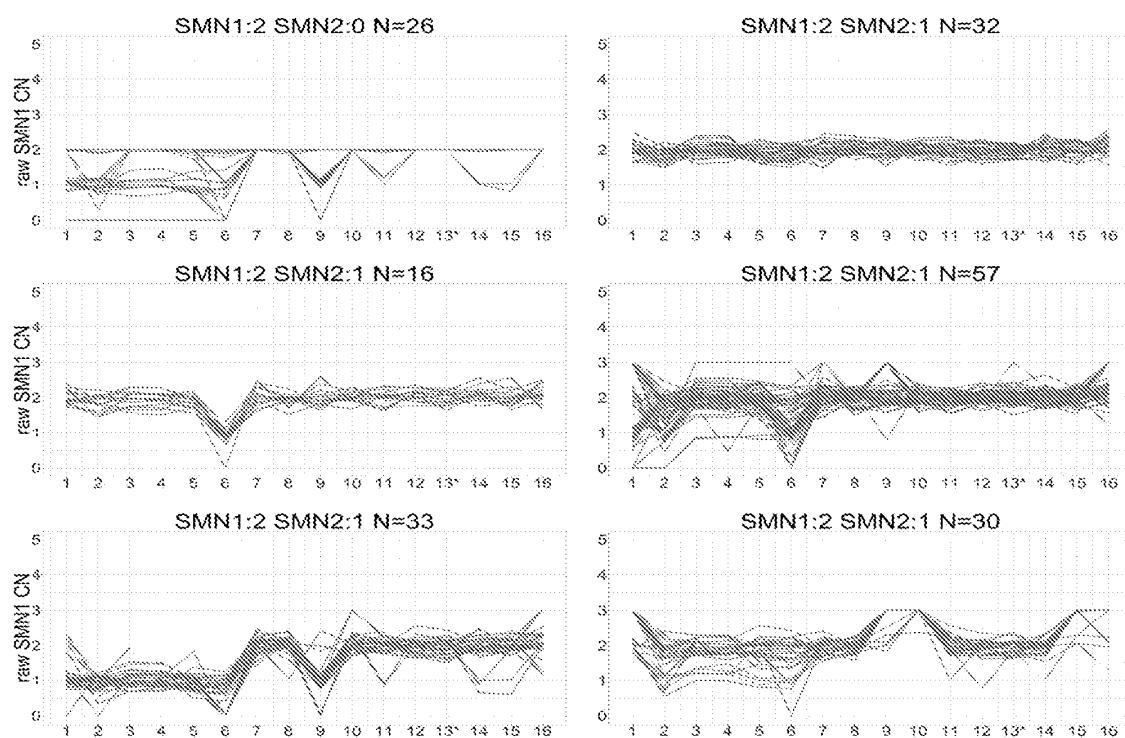
**FIG. 17**

FIG. 18A**FIG. 18B**

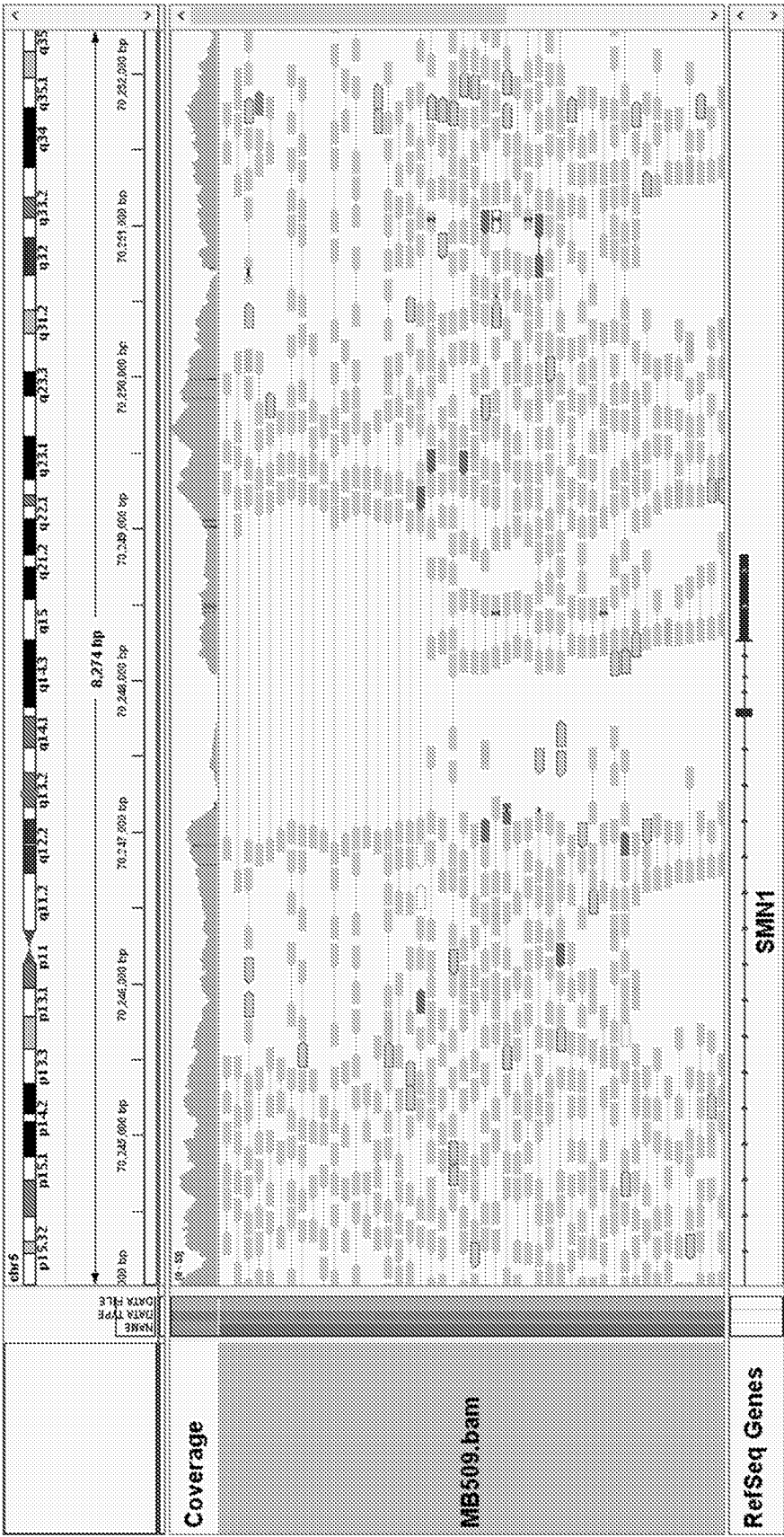


FIG. 19

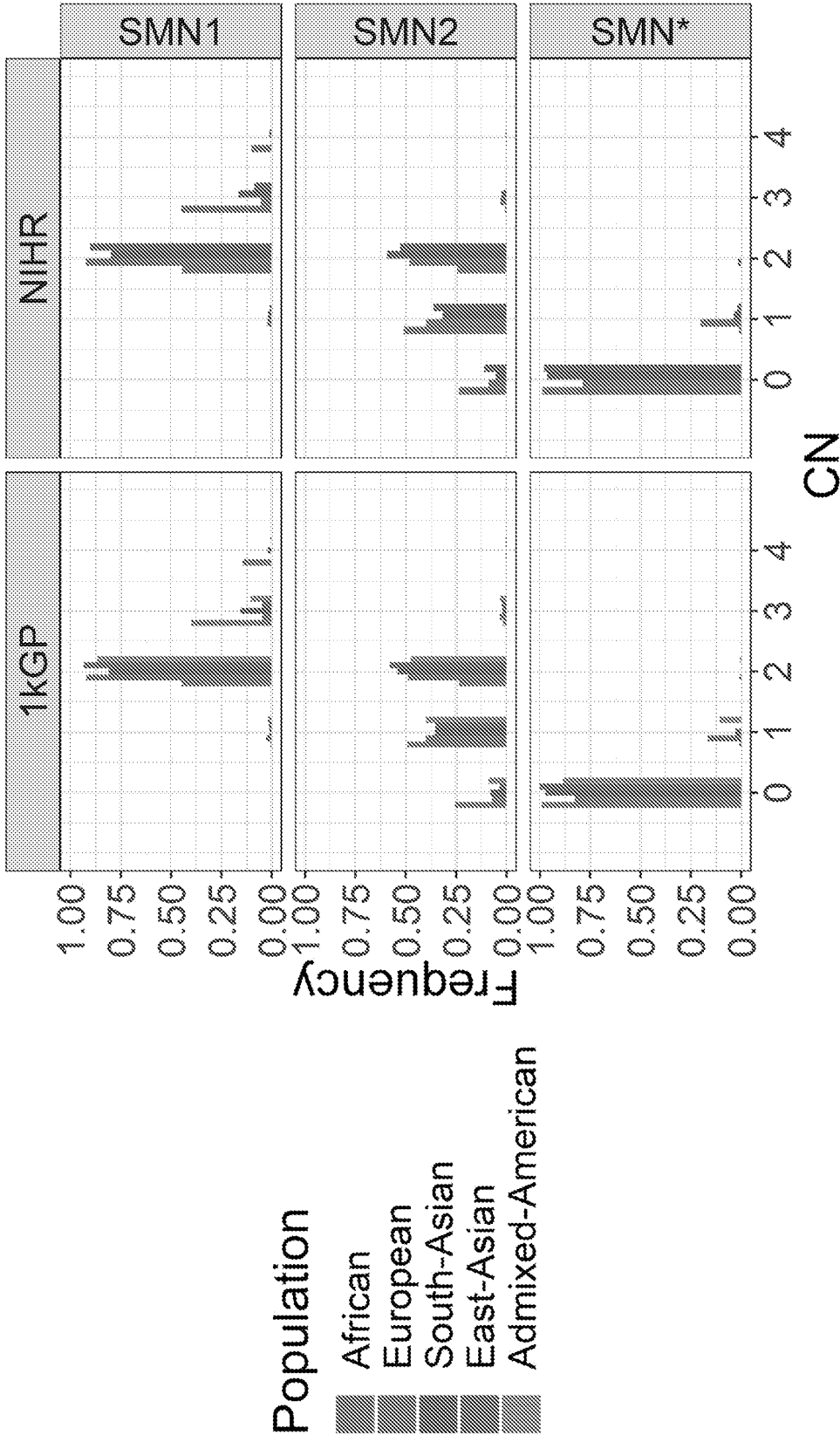


FIG. 20

FIG. 21A

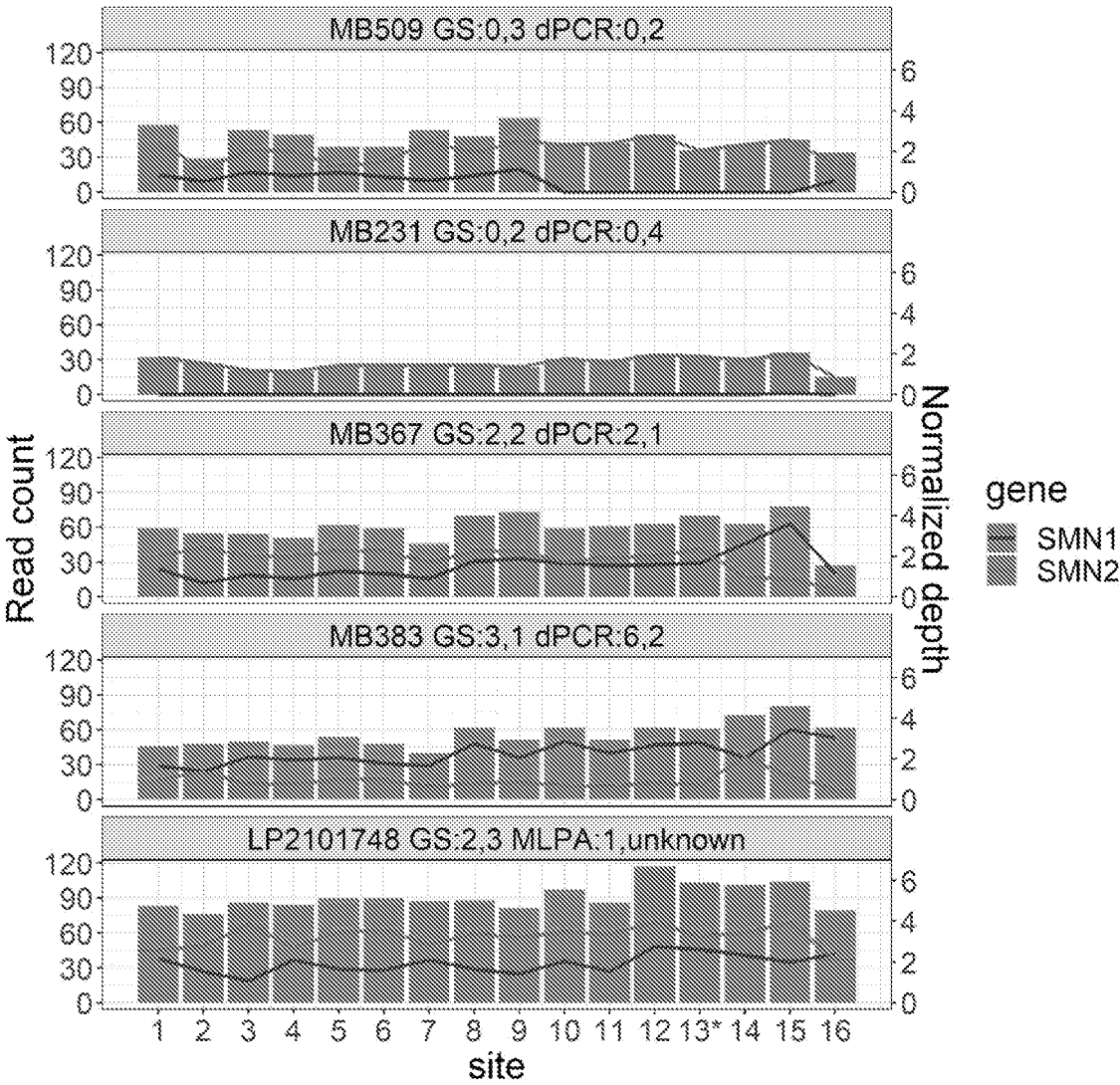
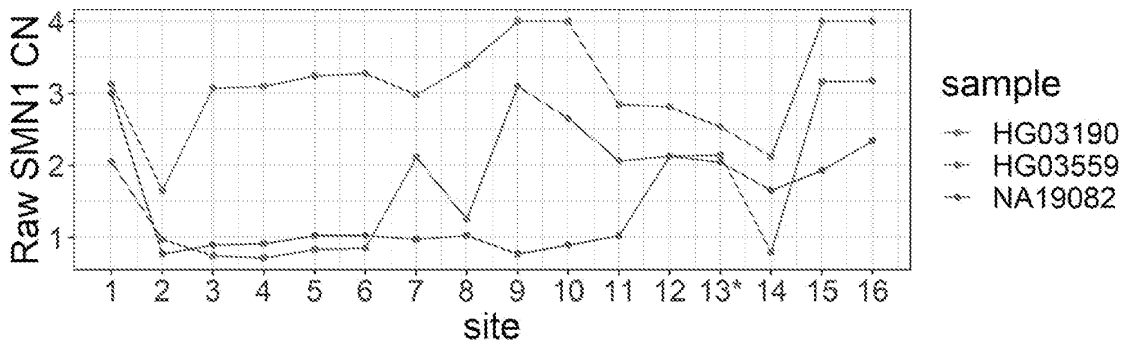


FIG. 21B



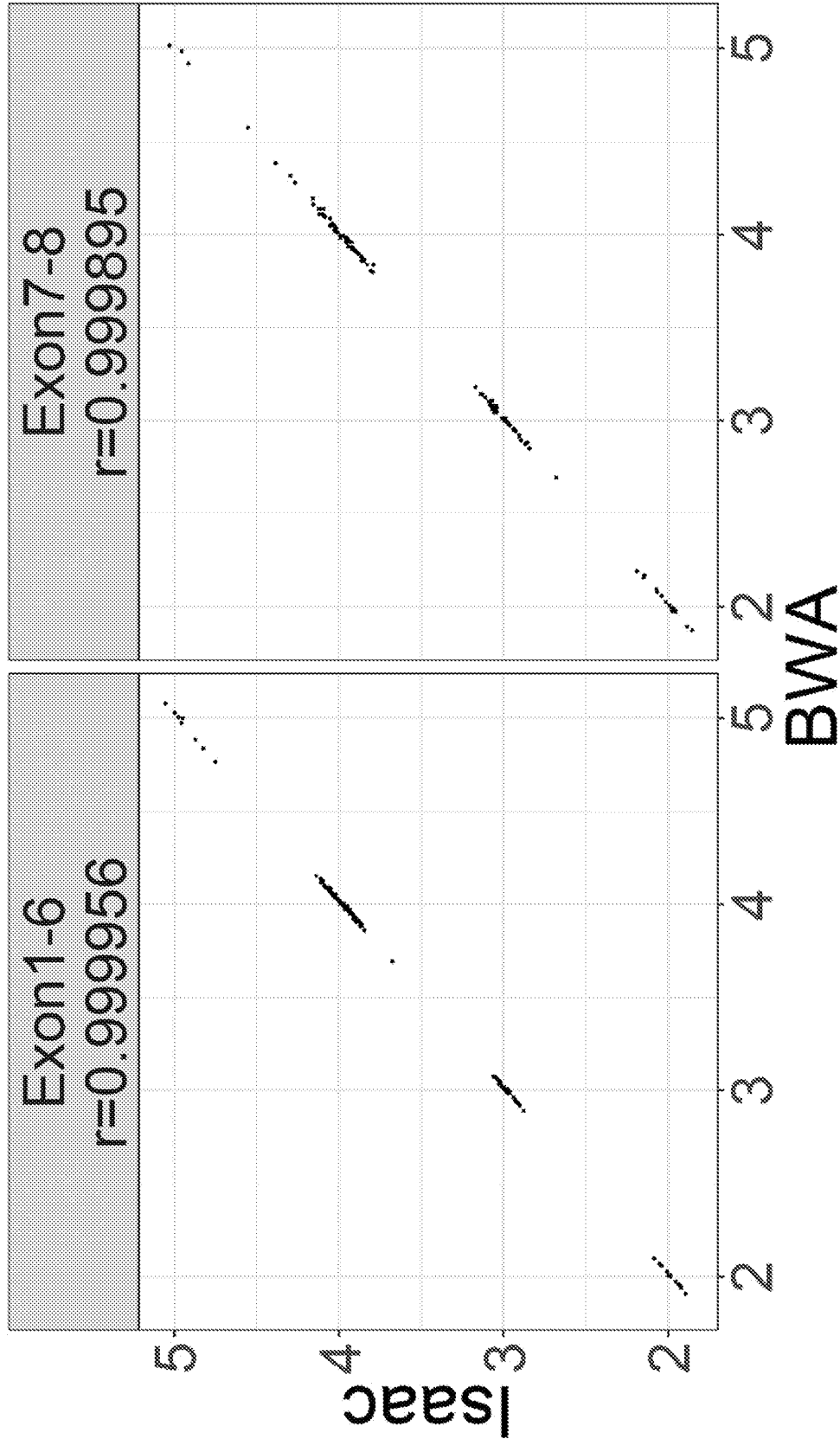


FIG. 22

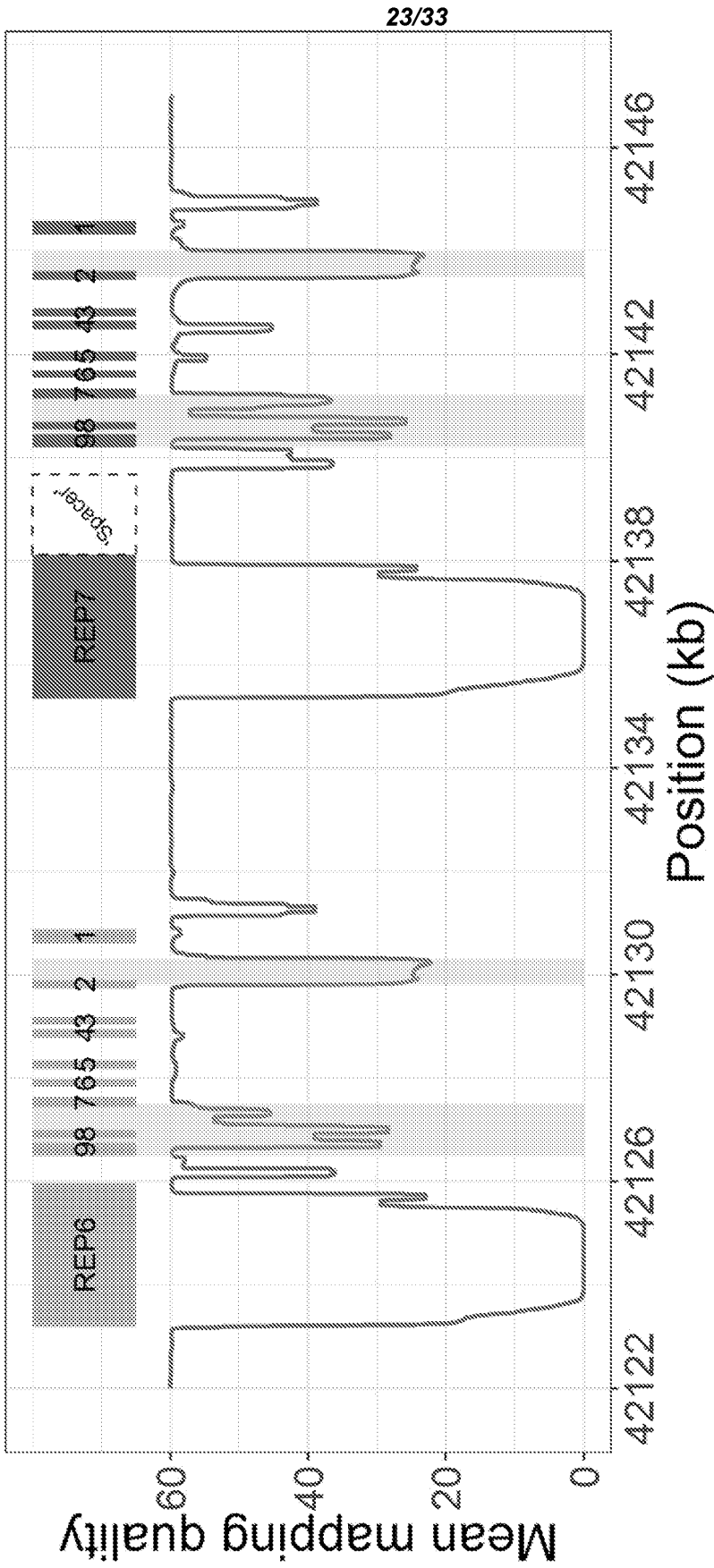


FIG. 23

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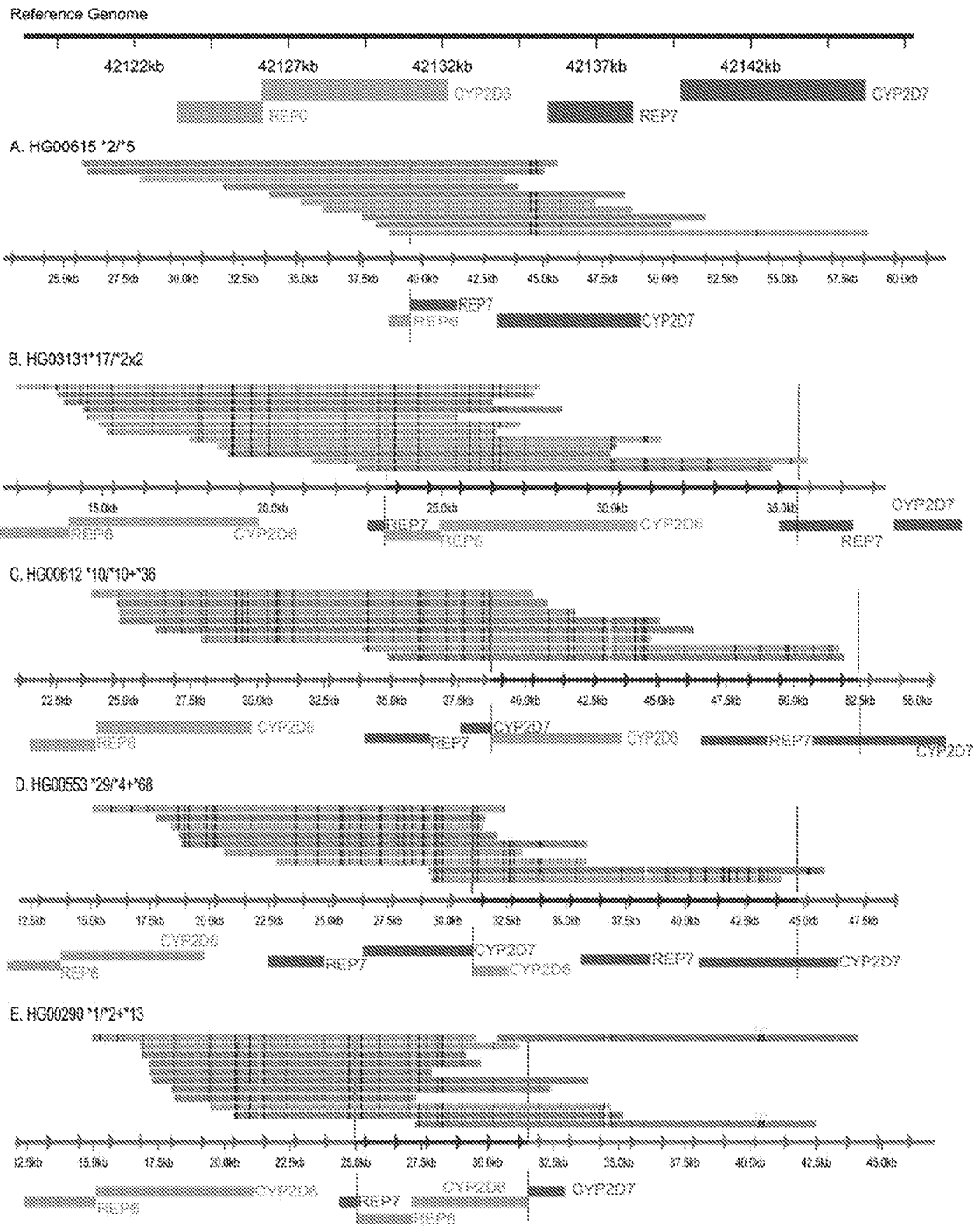


FIG. 24

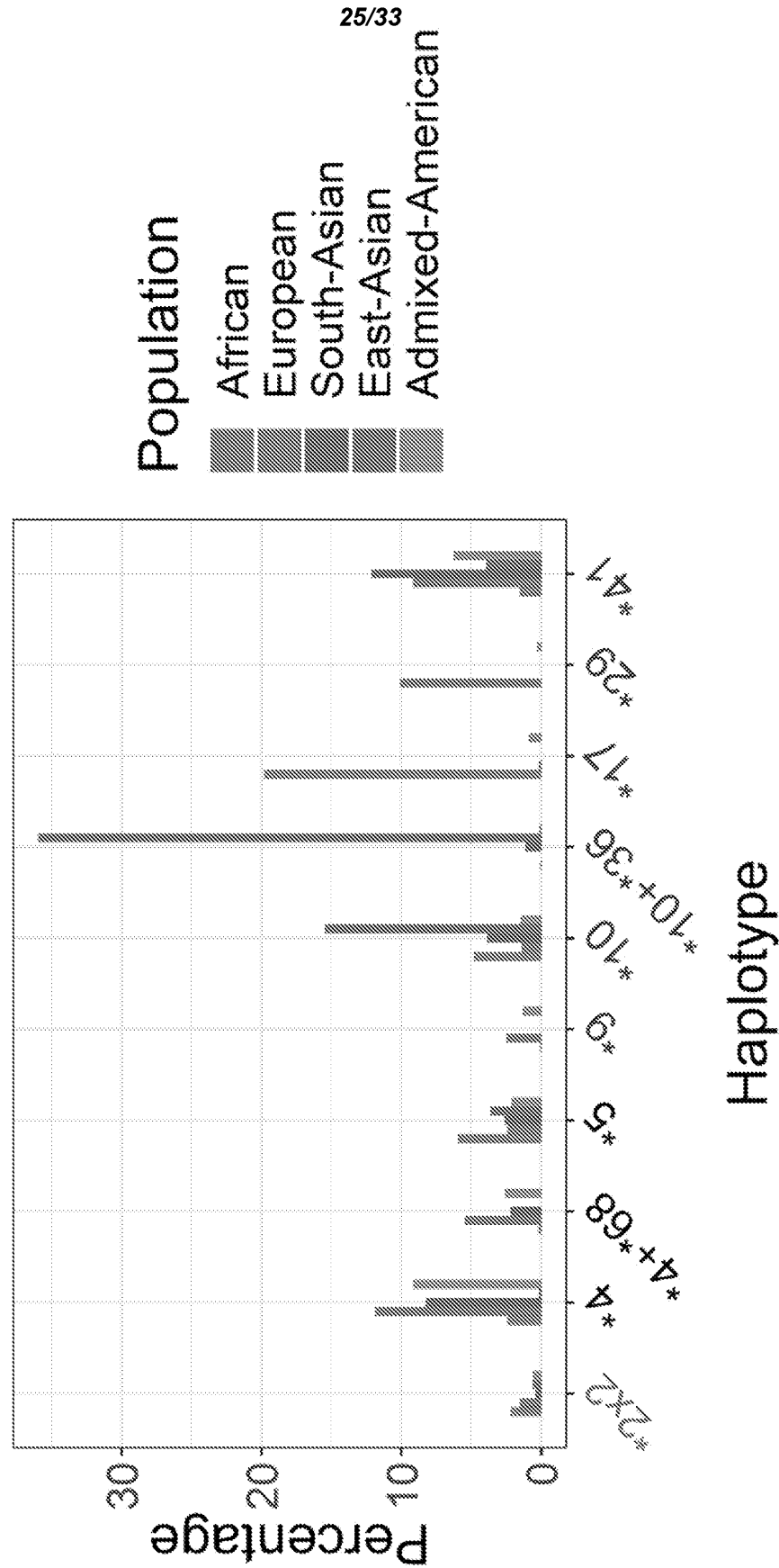


FIG. 25

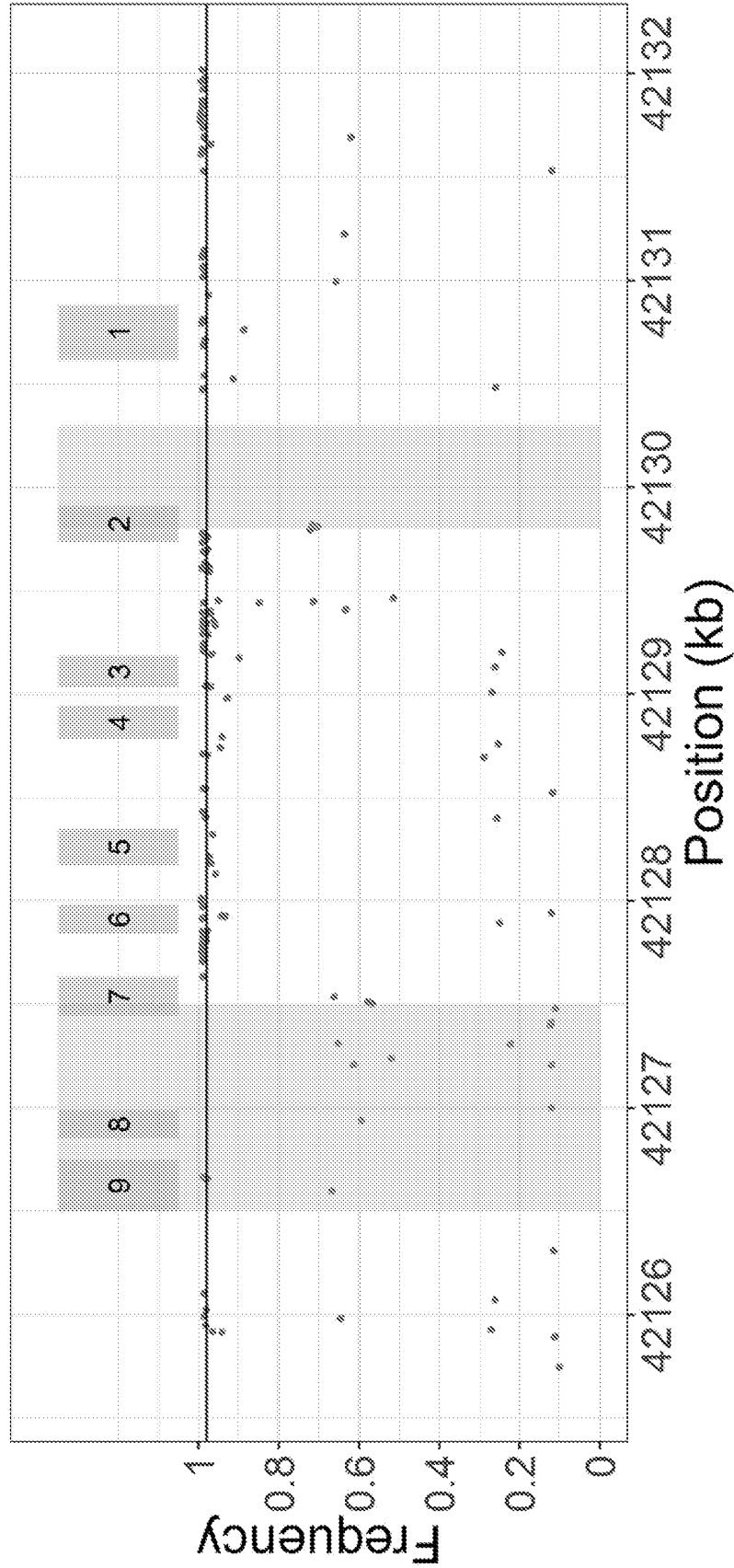


FIG. 26

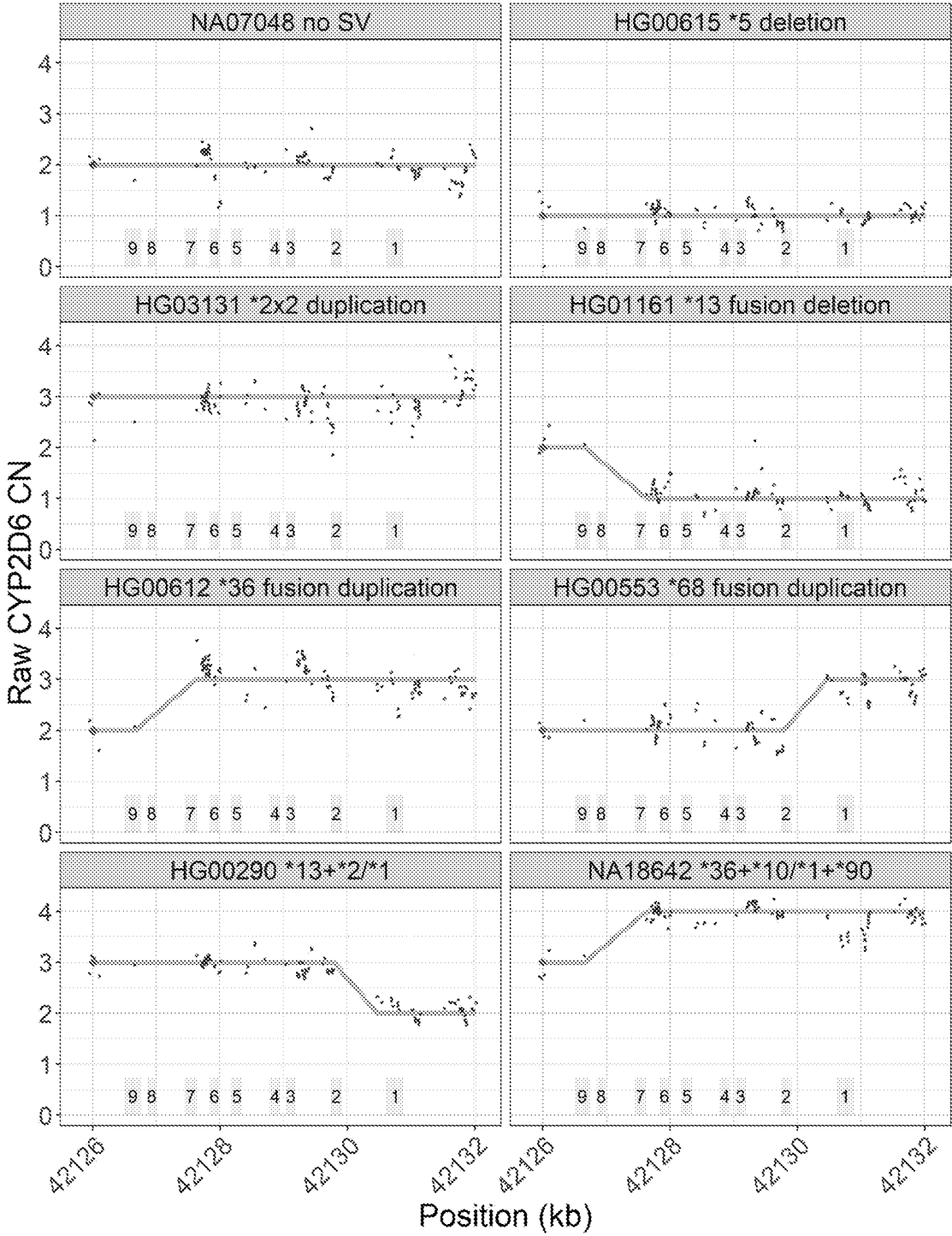


FIG. 27

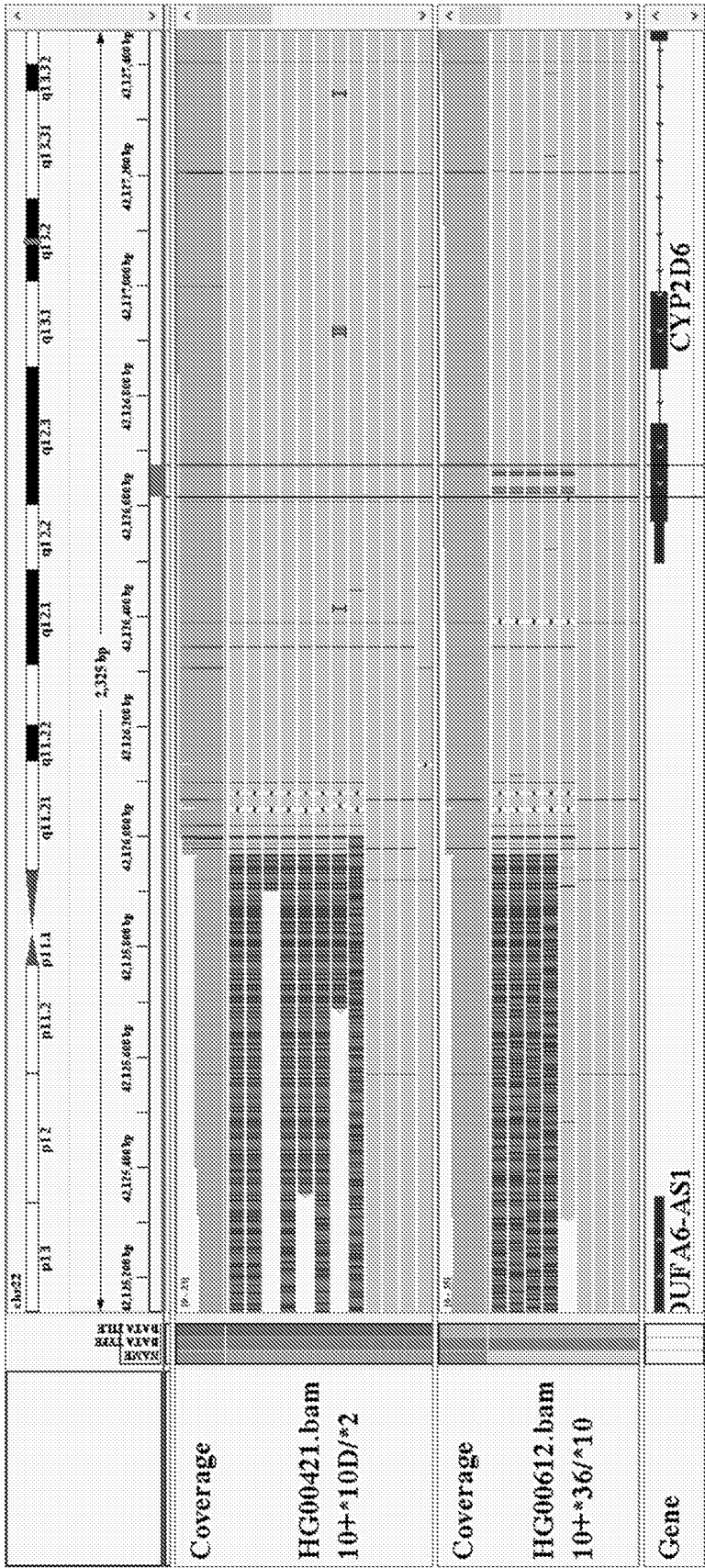


FIG. 28

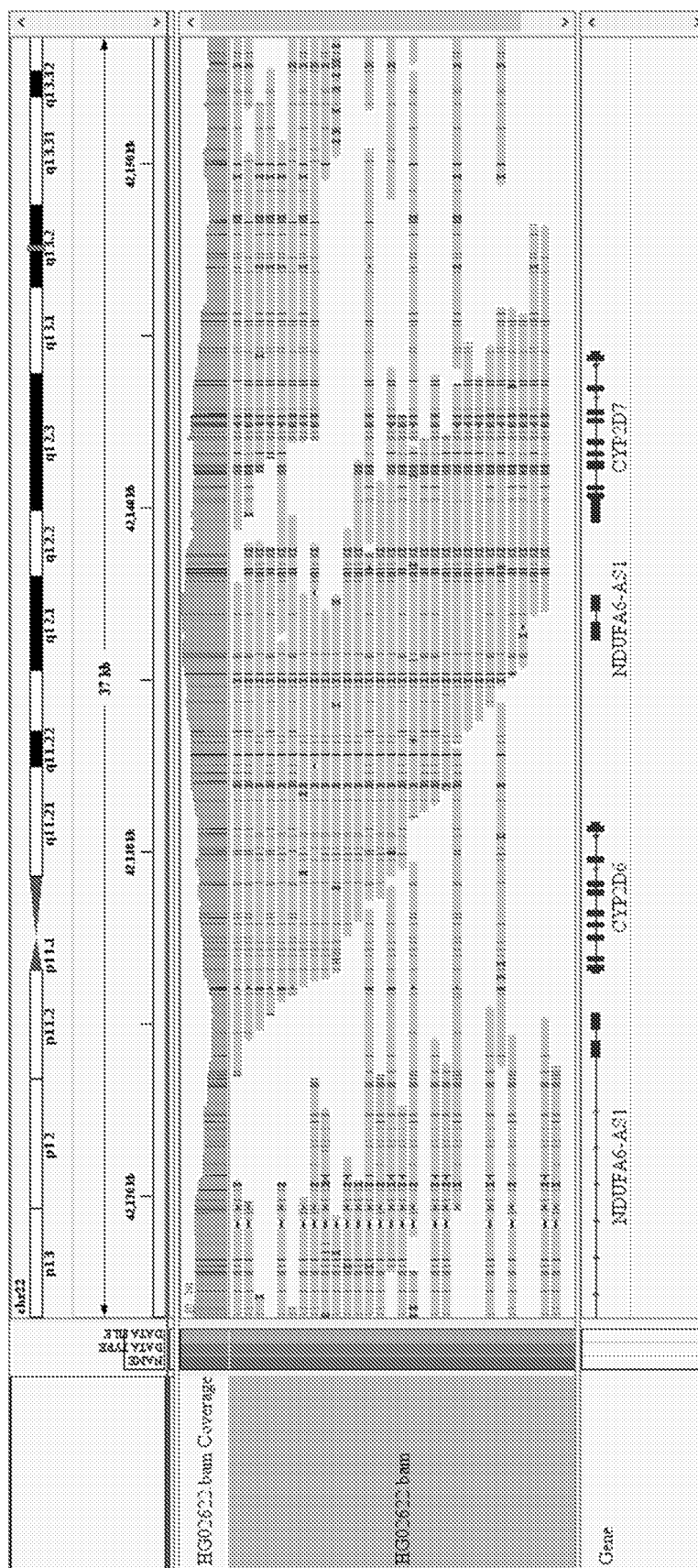


FIG. 29

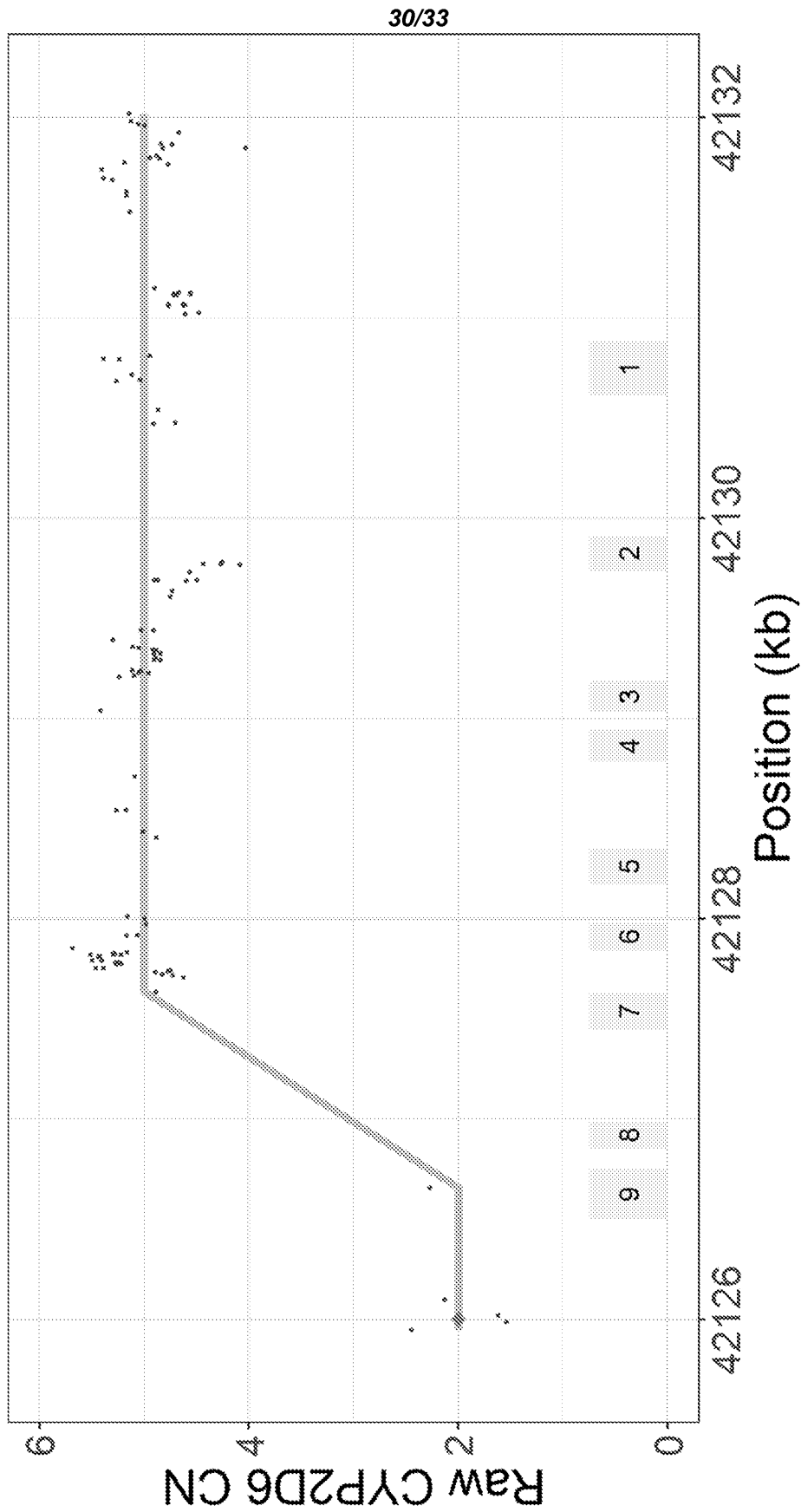


FIG. 30A

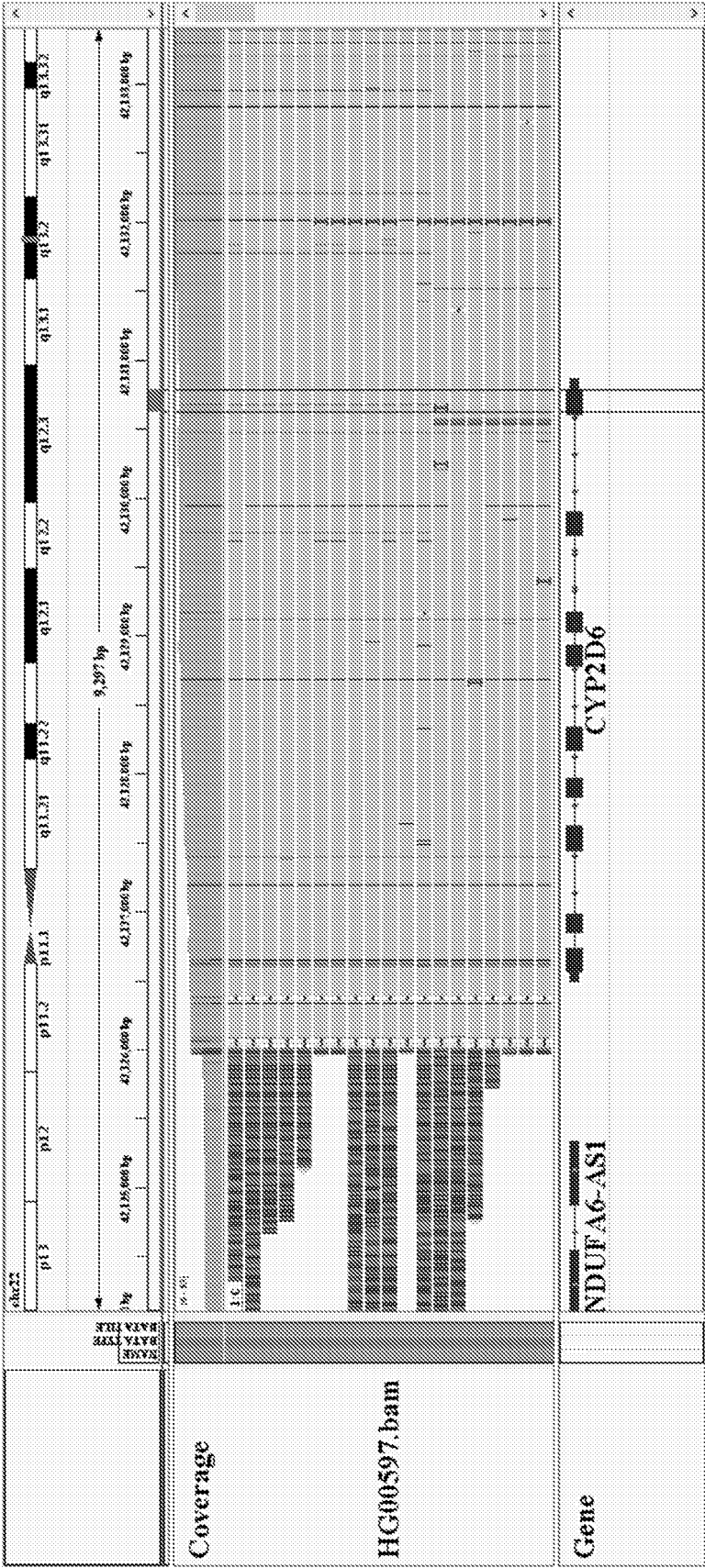


FIG. 30B

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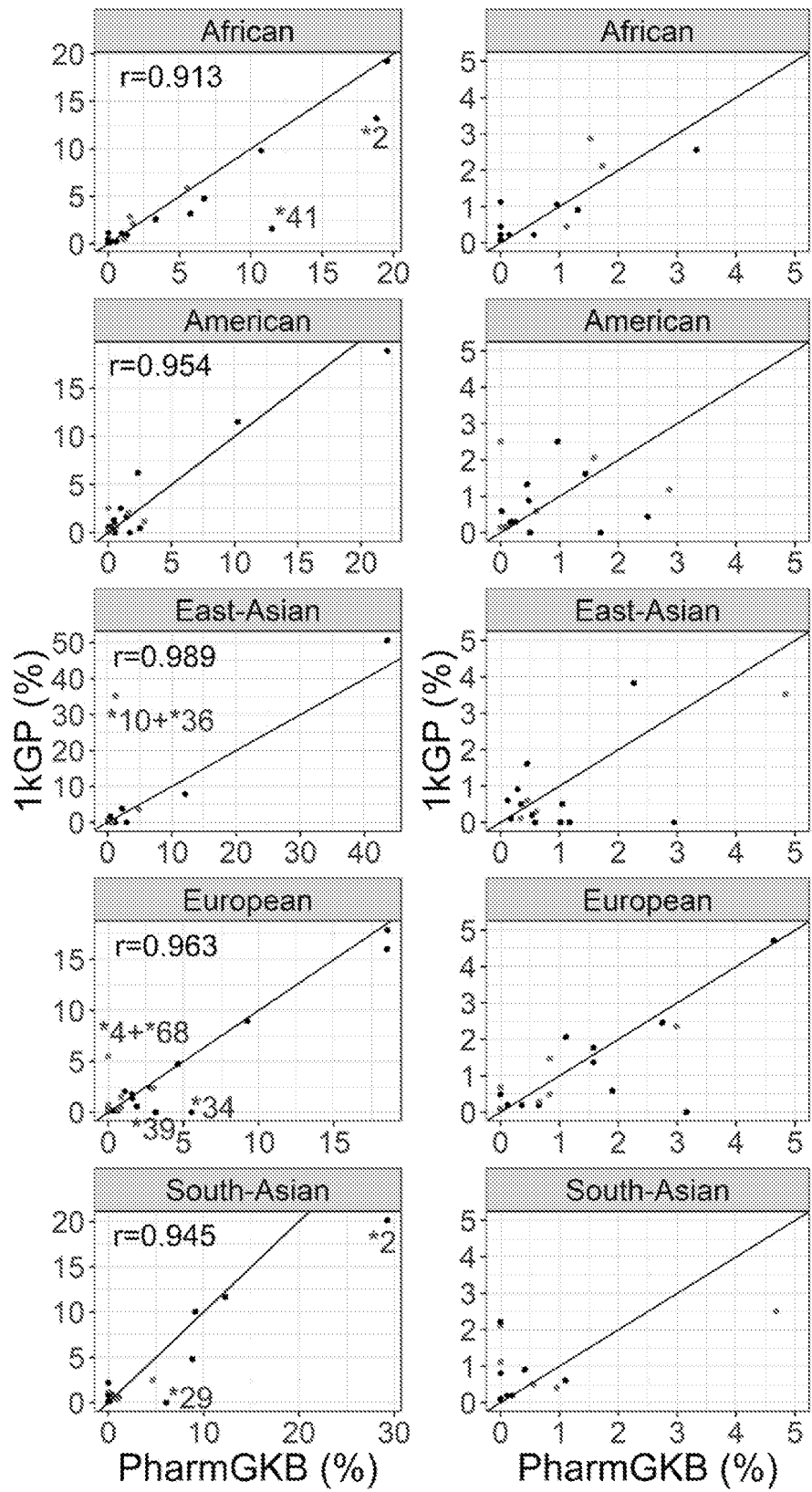


FIG. 31A

FIG. 31B

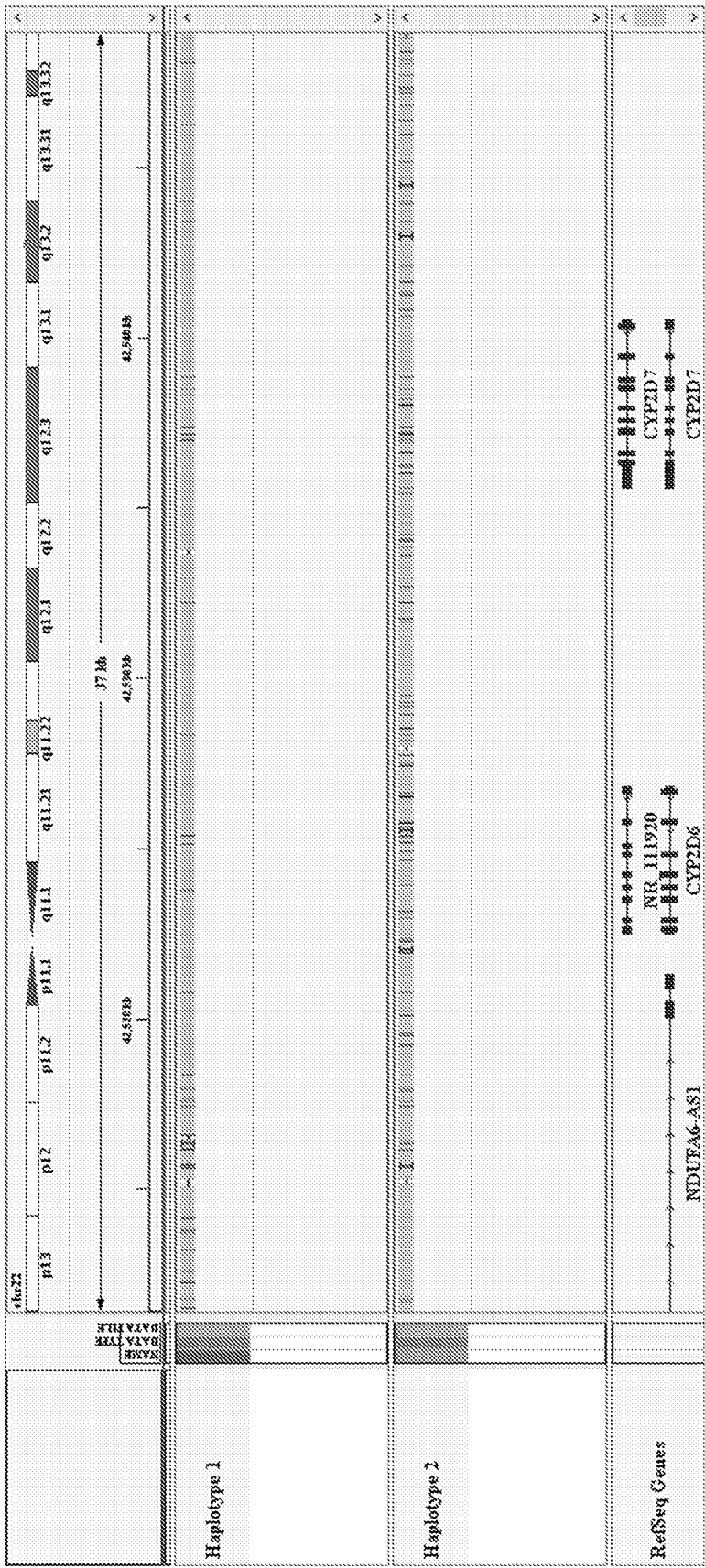


FIG. 32