

L1Hs call at chr1: 83144052

General information

L1Hs insertion: 1742bp L1Hs seq (-) 0/1

5' CAGAGCTGCAACAGGCCGAA AGCC **Insertion** AGCC CAGGTTGGGTGTTTCTG 3'

Empty site

5' CAGAGCTGCAACAGGCCGAA AGCC CAGGTTGGGTGTTTCTG 3'
3' GTCTCGACGTTGTCCGGCTT TCGG GTCCAACCCACAAAGAC 5'

Hallmarks

TSU length: 4bp
polyA length: 11bp
EN Cleavage site: 5' AGCC/C 3'

In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 6/32
Allele frequency: 9/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type2 (as missed by other callers, PALMER and PAV)

chr1:83,144,052-83,144,114



L1Hs call at chr2: 91993470

General information

L1Hs insertion: 836bp L1Hs seq (-) 0/1

5' GTCAGTTTATT AGTTTTCT **Insertion** AGTTTTCT AAAATAAAAAAA 3'

Empty site

5' GTCAGTTTATT AGTTTTCT AAAATAAAAAAA 3'
3' CAGTCAAATAA TCAAAAGA TTTTATTTTTTTT 5'

Hallmarks

TSU length: 8bp
polyA length: 35bp
EN Cleavage site: 5' TTCT/A 3'

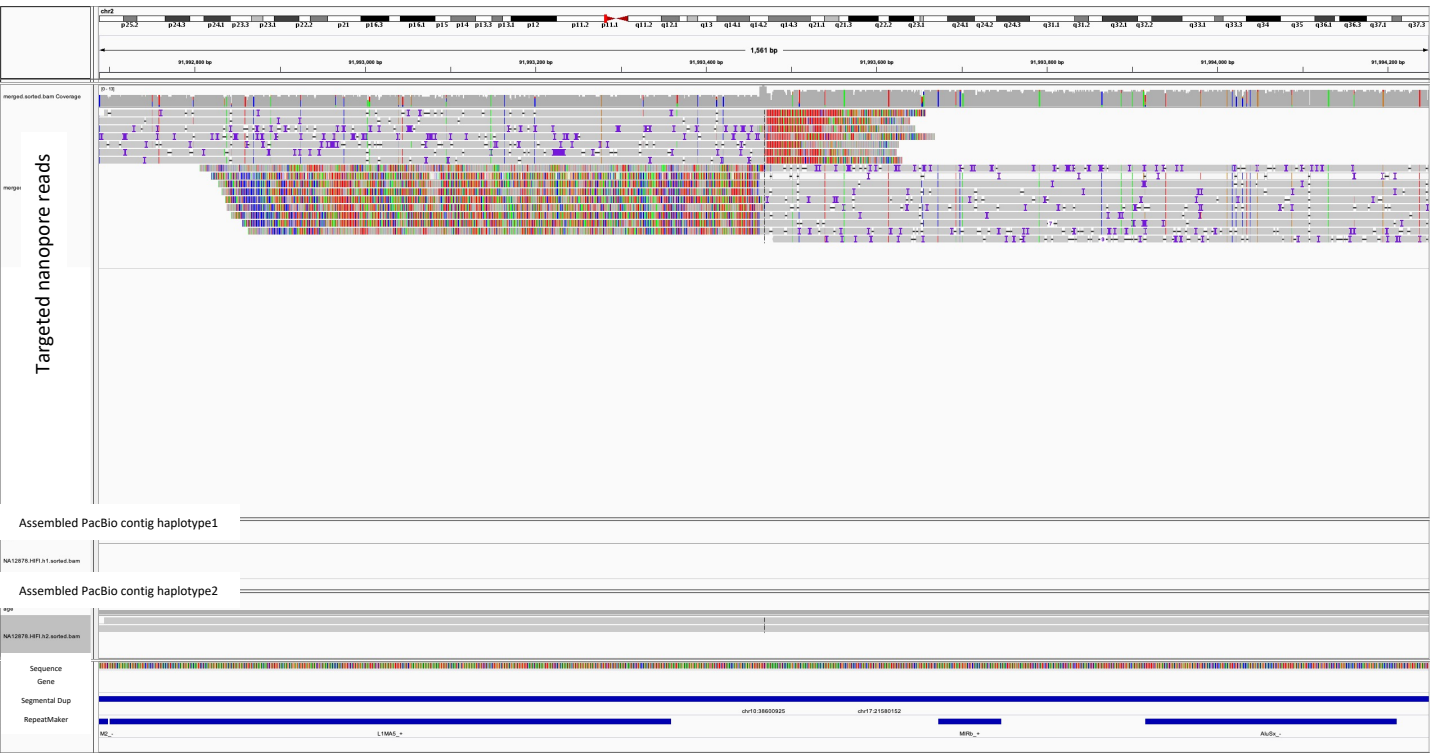
In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 7/32
Allele frequency: 7/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type1 (as missed by other technologies)

chr2:91,992,688-91,994,252



L1Hs call at chr2: 130095311

General information

L1Hs insertion: full-length L1Hs seq (-) 0/1

5' GTCAAACATTT CCATGTCAAACATTT **Insertion** CCATGTCAAACATTT TCAAAGTGAG 3'

Empty site

5' GTCAAACATTT CCATGTCAAACATTT TCAAAGTGAG 3'
3' CAGTTTGTAAAG GGTACAGTTTGTAAAG AGTTTCACTC 5'

Hallmarks

TSB length: 15bp
polyA length: 24bp
EN Cleavage site: 5' ATTT/T 3'

In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 4/32
Allele frequency: 4/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type1 (as missed by other technologies)

chr2:130,089,051-130,101,572



L1Hs call at chr2: 155671310

General information

L1Hs insertion: full-length L1Hs seq (+) 0/1

5' AAACCTCTGT CAAAAAAAAAAAAAGAAAGAAAGAAAGAA CAAAAAAAAAAAAAGAAAGAAAGAAAGAA AGAAATAAAA 3'

Insertion

Empty site

5' AAACCTCTGT CAAAAAAAAAAAAAGAAAGAAAGAAAGAA AGAAATAAAA 3'
3' TTTGAGACA GTTTTTTTTTTTTTCTTTCTTTCTTTCTTT TCTTTATTTT 5'

Hallmarks

TSB length: 28bp
polyA length: 9bp
EN Cleavage site: 5' TTTG/A 3'

In Ebert et al. 2021. Science

NA12878 genotype: 0/1 (mis-categorized)
Sample frequency: 12/32
Allele frequency: 16/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type2 (as missed or mis-categorized by other callers, PALMER and PAV)

chr2:155,665,049-155,677,570



AluY call at chr2: 242181271

General information

L1Hs insertion: 190bp AluY seq (+) 0/1

5' TTCTCTTG AGATGCCCCAATC **Insertion** AGATGCCCCAATC ATCATT 3'

Empty site

5' TTCTCTTG AGATGCCCCAATC ATCATT 3'
3' AAGAGAAC TCTACGGGGTTAG TAGTAA 5'

Hallmarks

TSU length: 13bp
polyA length: 39bp
EN Cleavage site: 5' ATCT/C 3'

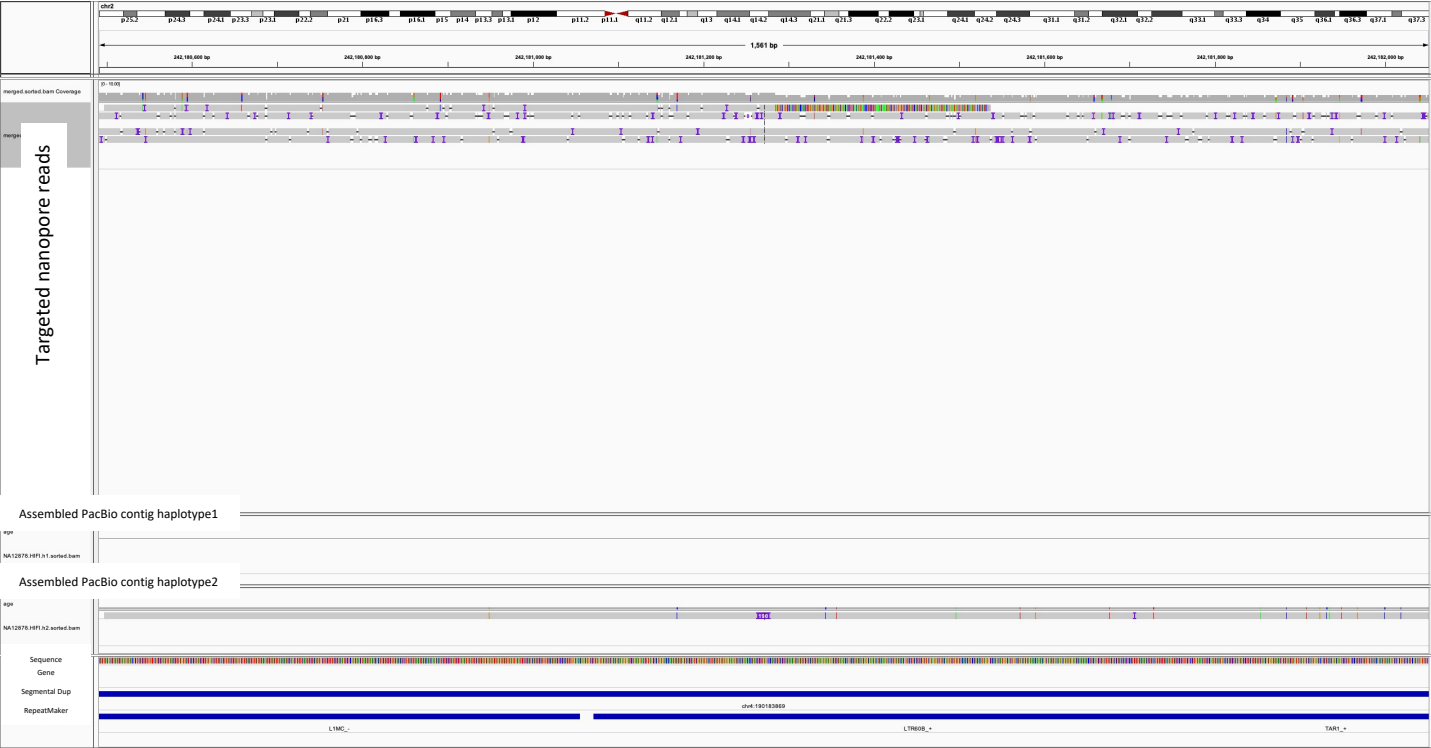
In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 14/32
Allele frequency: 14/64

IGV screenshot (general information can not be defined by nanopore reads)

True positive type2 (as missed by other callers, PALMER and PAV)

chr2:242,180,491-242,182,055



AluY call at chr5: 17712479

General information

L1Hs insertion: 190bp AluY seq (+) 0/1

5' TTTATCTCTT AGAAAAATATACA **Insertion** AGAAAAATATACA CAAGAAAT 3'

Empty site

5' TTTATCTCTT AGAAAAATATACA CAAGAAAT 3'
3' AAATAGAGAA TCTTTTATATGT GTTCTTTA 5'

Hallmarks

TS length: 13bp
polyA length: 22bp
EN Cleavage site: 5' TTCT/A 3'

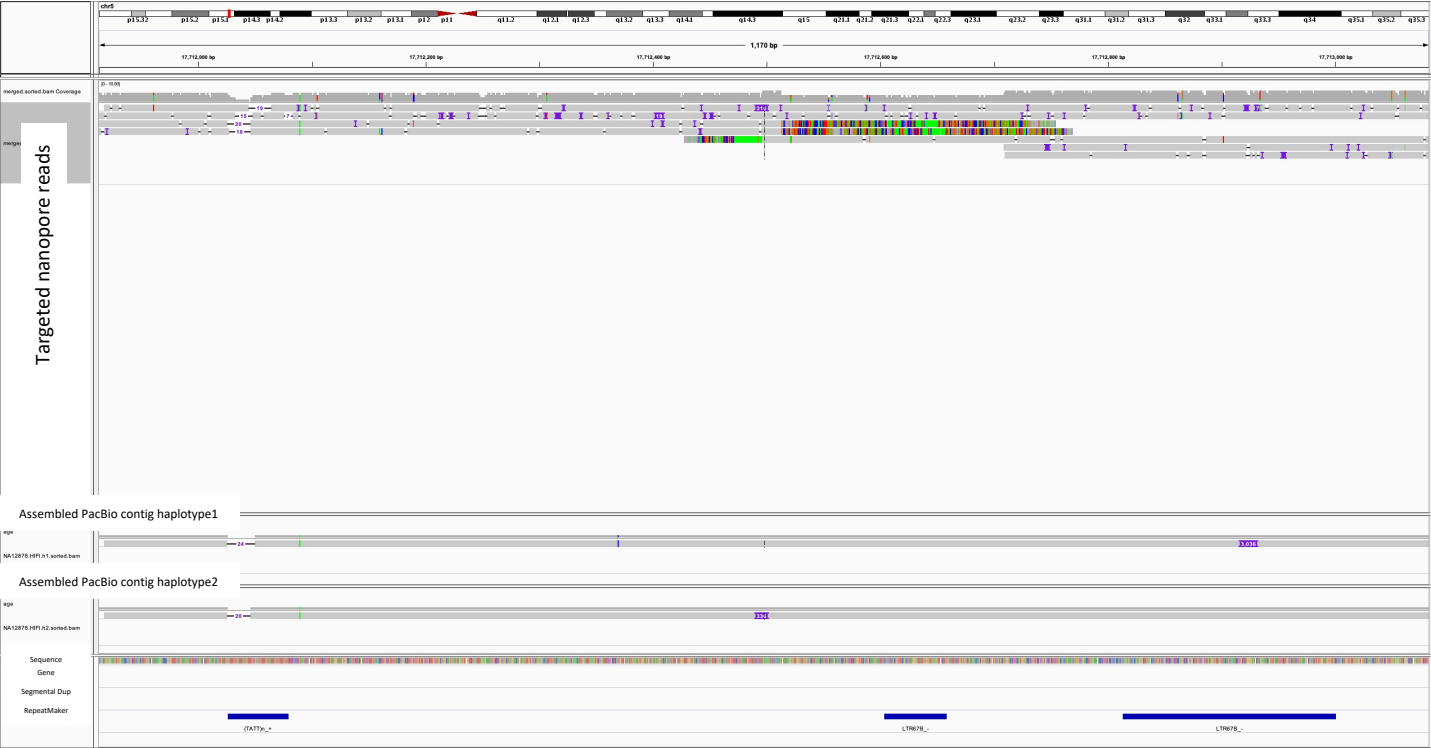
In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 28/32
Allele frequency: 39/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type2 (as missed by other callers, PALMER and PAV)

chr5:17,711,913-17,713,085





L1Hs call at chr6: 126302449

General information

L1Hs insertion: 389bp L1Hs seq (-) 0/1

5' ACTGACTGCCTT TGTT Insertion TGTT AATTGTCTCTC 3'

Empty site

5' ACTGACTGCCTT TGTT AATTGTCTCTC 3'
3' TGA CTGACGAAA ACAA TTAACAGAGAG 5'

Hallmarks

TSU length: 4bp
polyA length: 74bp
EN Cleavage site: 5' TGTT/A 3'

In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 2/32
Allele frequency: 2/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type2 (as missed by other callers, PALMER and PAV)

chr6:126,302,174-126,302,724



L1Hs call at chr10: 4997072

General information

L1Hs insertion: 420bp L1Hs seq (-) 0/1

5' TGACCACTGT TAGCTTAACCAA **Insertion** TAGCTTAACCAA GTTTTTTAAA 3'

Empty site

5' TGACCACTGT TAGCTTAACCAA GTTTTTTAAA 3'
3' ACTGGTGACA ATCGAATTGGTT CAAAAATTT 5'

Hallmarks

TSU length: 12bp
polyA length: 14bp
EN Cleavage site: 5' CCAA/G 3'

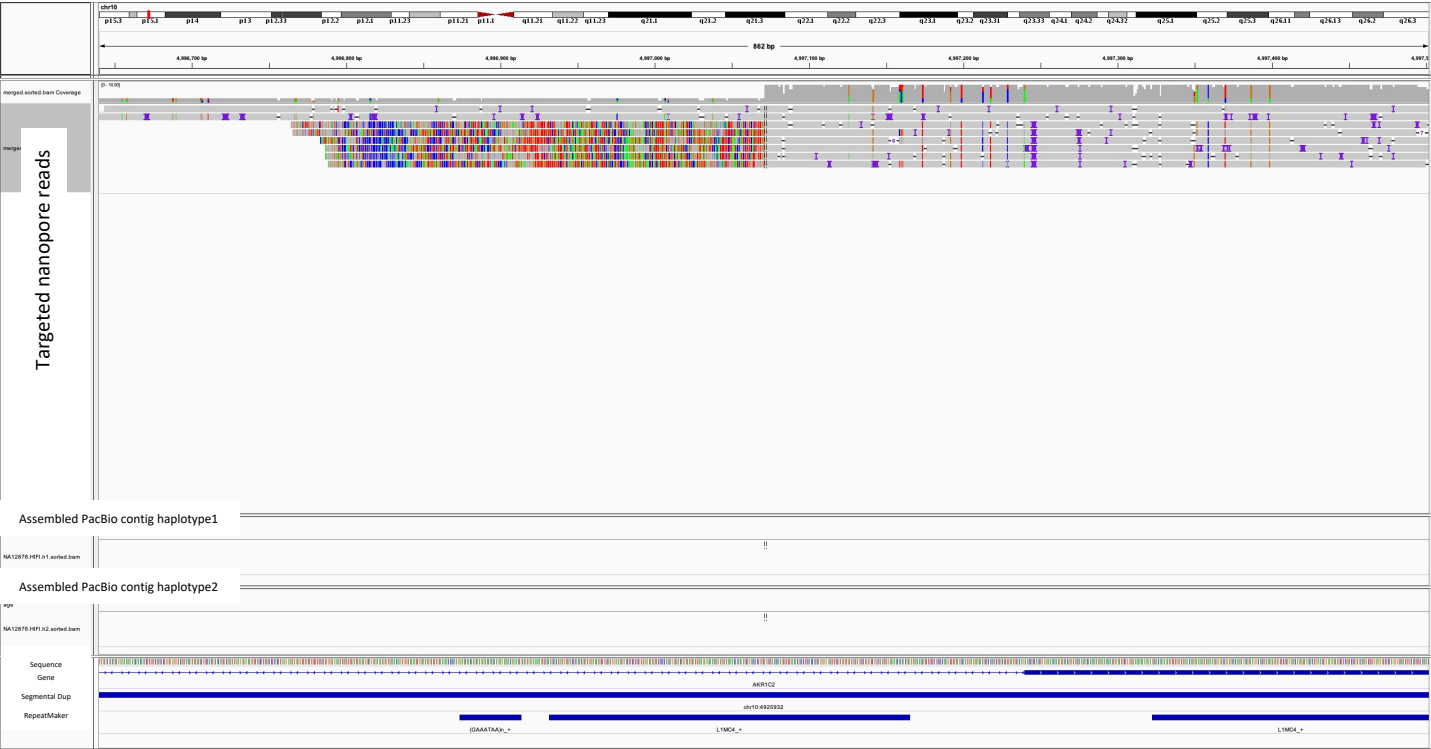
In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 4/32
Allele frequency: 5/64

IGV screenshot (general information can not be defined by nanopore reads)

True positive type1 (as missed by other technologies)

chr10:4,996,640-4,997,503



L1Hs call at chr10: 38748664

General information

L1Hs insertion: 823bp L1Hs seq (-) 0/1

5' TCAGTTTATT AGTTTCT **Insertion** AGTTTCT AAAATAAAAAAAAA 3'

Empty site

5' TCAGTTTATT AGTTTCT AAAATAAAAAAAAA 3'
3' AGTCAAATAA TCAAAAGA TTTTATTTTTTTTT 5'

Hallmarks

TSU length: 8bp
polyA length: 75bp
EN Cleavage site: 5' TTCT/A 3'

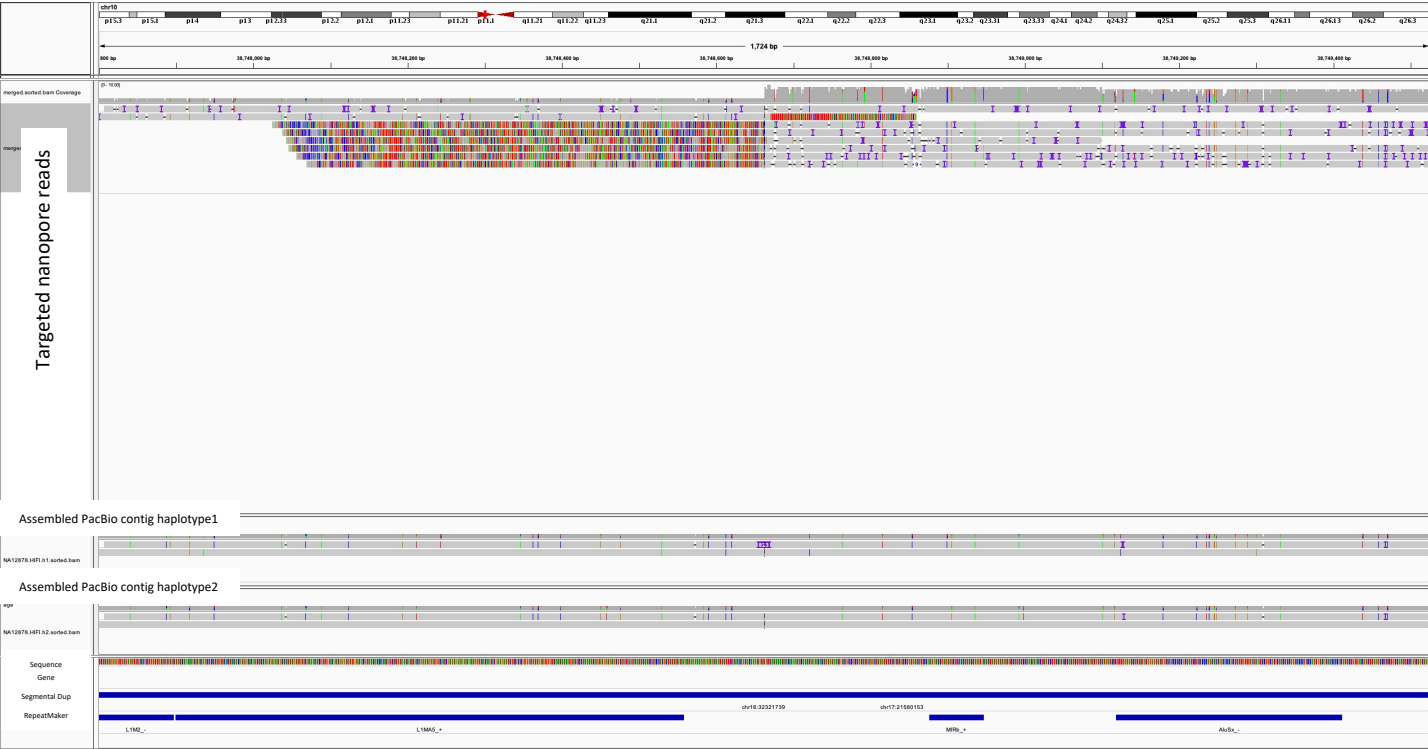
In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 0/32
Allele frequency: 0/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type2 (as missed by other callers, PALMER and PAV)

chr10:38,747,800-38,749,528



AluY call at chr16: 69157891

General information

L1Hs insertion: 323bp AluY seq (-) 0/1

5' CTGTGCCTGGC CCAAAAGTATTC **Insertion** CCAAAAGTATTC TTATACAT 3'

Empty site

5' CTGTGCCTGGC CCAAAAGTATTC TTATACAT 3'
3' TACACGGACCG GGTTTTTCATAAG AATATGTA 5'

Hallmarks

TS length: 12bp
polyA length: 26bp
EN Cleavage site: 5' ATTC/T 3'

In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 9/32
Allele frequency: 10/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type2 (as missed by other callers, PALMER and PAV)

chr16:69,157,321-69,158,487



L1Hs call at chr17: 18755862

General information

L1Hs insertion: 1202bp L1Hs seq (-) 0/1

5' TTAAGAACCT TGACCCAACATTTTCT **Insertion** TGACCCAACATTTTCT AACTCATGG 3'

Empty site

5' TTAAGAACCT TGACCCAACATTTTCT AACTCATGG 3'
3' AATTC'TTGGA ACTGGG'TGTAAAAGA TTGAGTACC 5'

Hallmarks

TSU length: 16bp
polyA length: 20bp
EN Cleavage site: 5' TTCT/A 3'

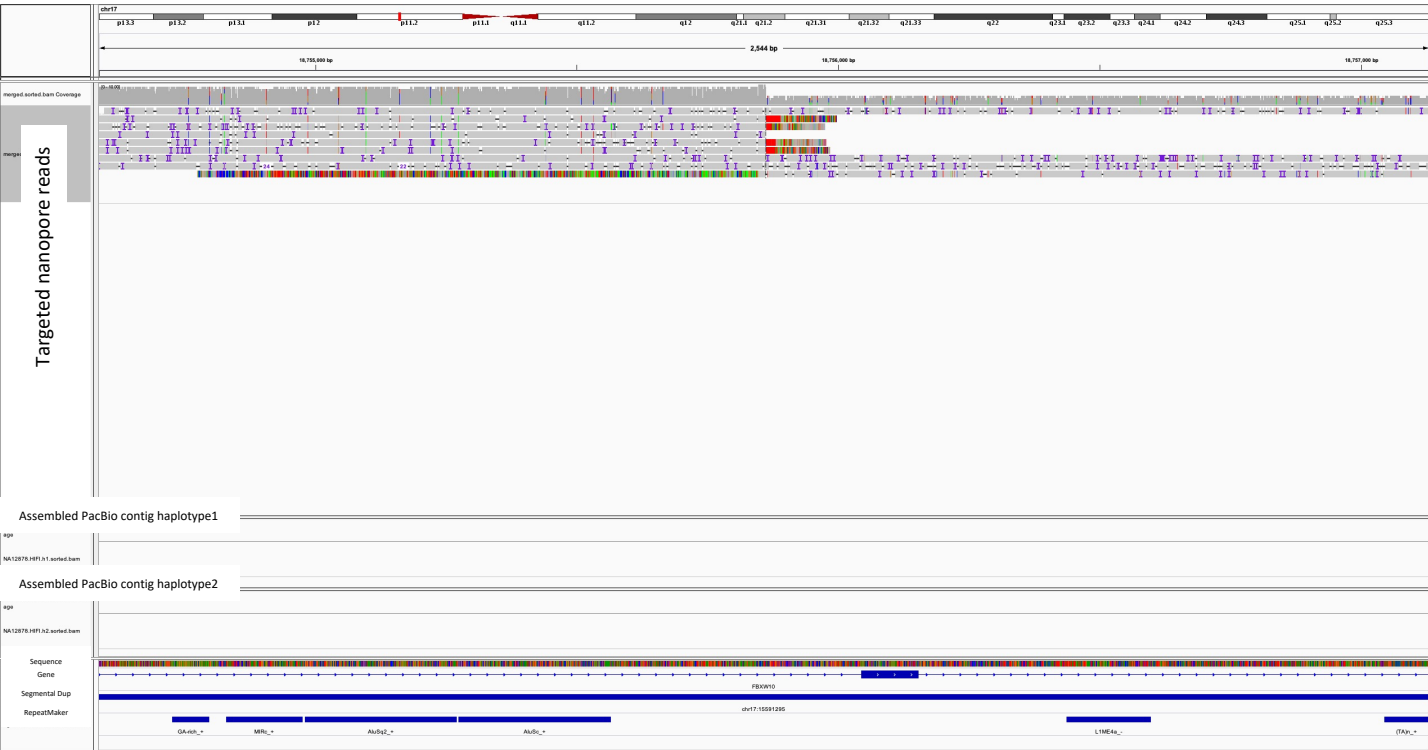
In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 1/32
Allele frequency: 1/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type1 (as missed by other technologies)

chr17:18,754,586-18,757,137



L1Hs call at chr17: 21828248

General information

L1Hs insertion: 1300bp L1Hs seq (+) 0/1 (with a 700bp 5' inverted L1 segment)

5' TTAATTCTATT GAAAAA **Insertion** GAAAAA CCAAATCTGTAGGAA 3'

Empty site

5' TTAATTCTATT GAAAAA CCAAATCTGTAGGAA 3'
3' AATTAAGATAA CTTTTT GGTTTAGACATCCTT 5'

Hallmarks

TSU length: 6bp
polyA length: 29bp
EN Cleavage site: 5' TTCT/A 3'

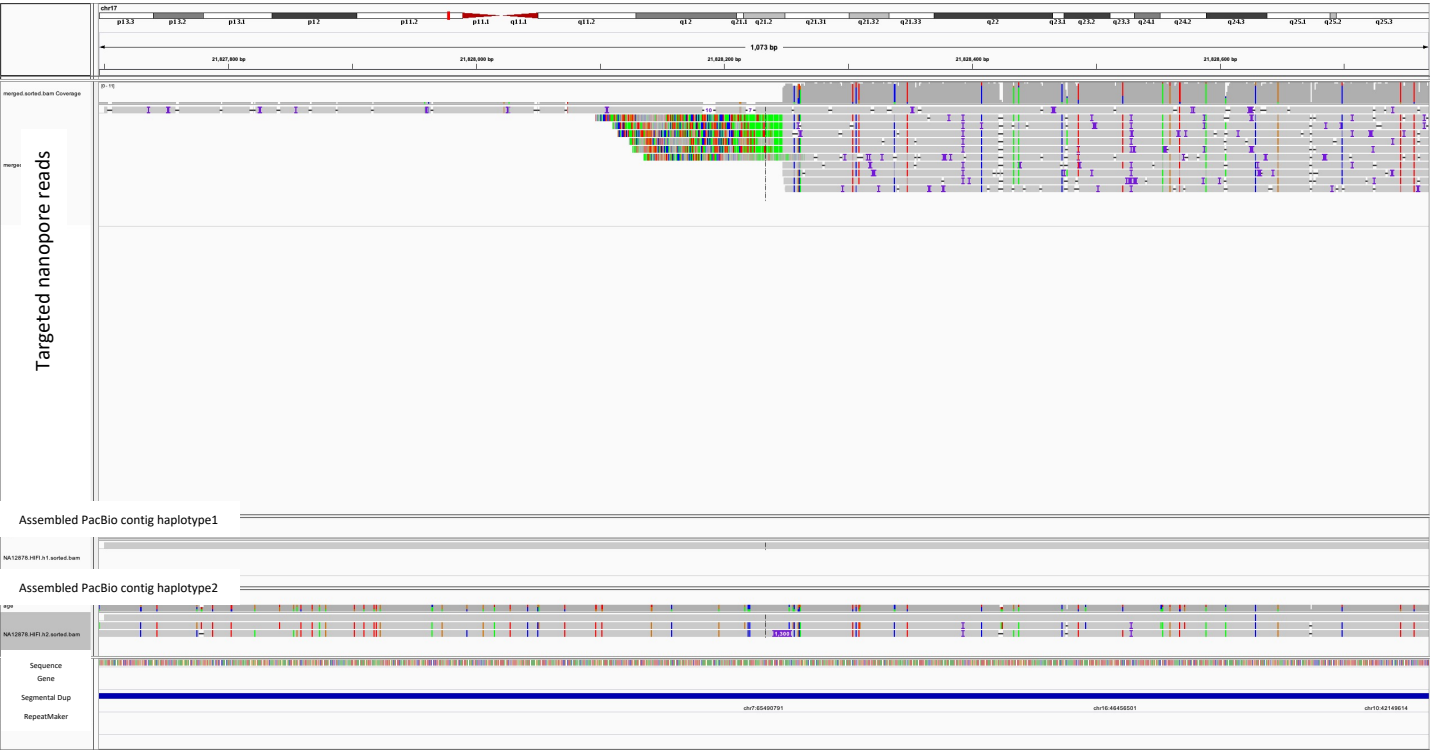
In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 1/32
Allele frequency: 1/64

IGV screenshot (general information can not be defined by nanopore reads)

True positive type2 (as missed by other callers, PALMER and PAV)

chr17:21,827,696-21,828,771



AluY call at chr19: 52384798

General information

L1Hs insertion: 467bp AluY seq (+) 1/1 (with a 158bp 3' end segment)

5' ACGGGAGAGCA ACCTTACAAATGTAATGAATTTGG **Insertion** ACCTTACAAATGTAATGAATGTGG CAAAGCAT 3'

Empty site

5' ACGGGAGAGCA ACCTTACAAATGTAATGAATGTGG CAAAGCAT 3'
3' TGCCCTCTCGT TGGAATGTTTACATTACTTACACC GTTTCGTA 5'

Hallmarks

TS length: 24bp
polyA length: 39bp
EN Cleavage site: 5' AGGT/T 3'

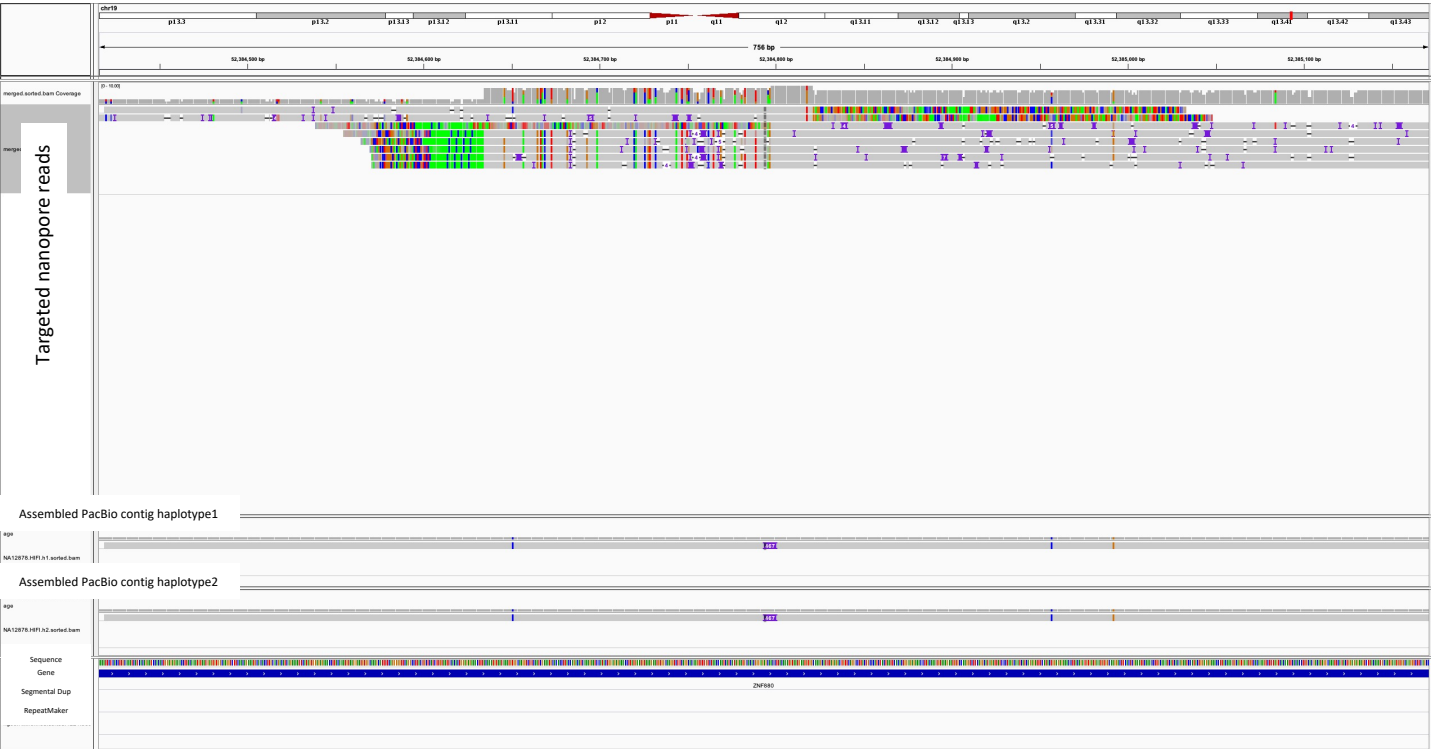
In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 9/32
Allele frequency: 10/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type2 (as missed by other callers, PALMER and PAV)

chr19:52,384,416-52,385,172



AluY call at chr19: 58605628

General information

L1Hs insertion: 322bp AluY seq (+) 0/1

5' GAACTTCTCT TAAGGTGCCCCAATC **Insertion** TGAGATGCCCCAATC ATCATTCACT 3'

Empty site

5' GAACTTCTCT TAAGGTGCCCCAATC ATCATTCACT 3'
3' CTTGAAGAGA ATTCCACGGGGTTAG TAGTAAGTGA 5'

Hallmarks

TSU length: 15bp
polyA length: 42bp
EN Cleavage site: 5' CTTA/A 3'

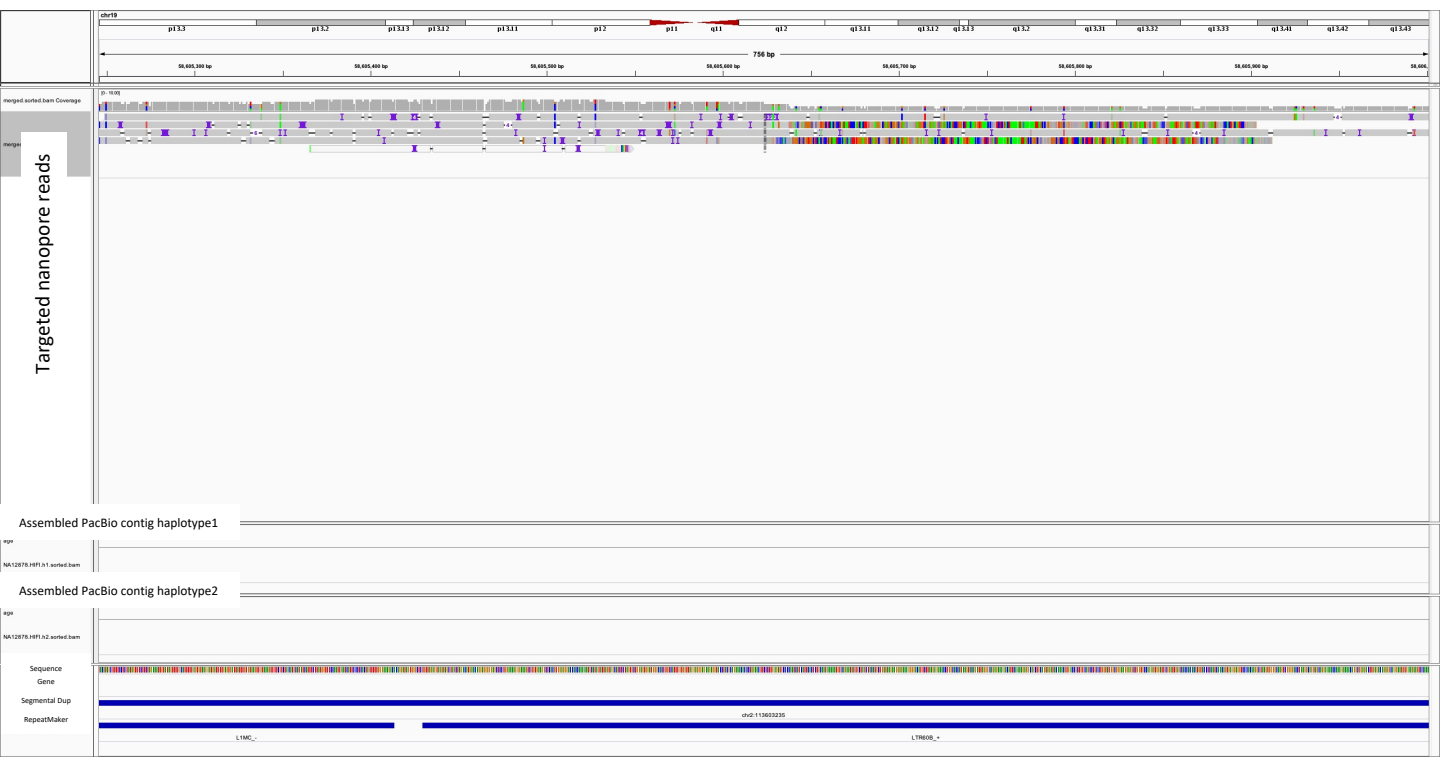
In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 10/32
Allele frequency: 13/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type2 (as missed by other callers, PALMER and PAV)

chr19:58,605,246-58,606,002



L1Hs call at chr20: 30176989

General information

L1Hs insertion: 1011bp L1Hs seq (-) 0/1 (with 228bp 5' inverted L1 segment)

5' tgtgttaggaa aattccacatctttc **Insertion** aattccacatctttc ctctagccatt 3'

Empty site

5' tgtgttaggaa aattccacatctttc ctctagccatt 3'
3' acacaatcctt ttaagggtgtagaag gagatcggtaa 5'

Hallmarks

TSB length: 15bp
polyA length: 16bp
EN Cleavage site: 5' TTTC/C 3'

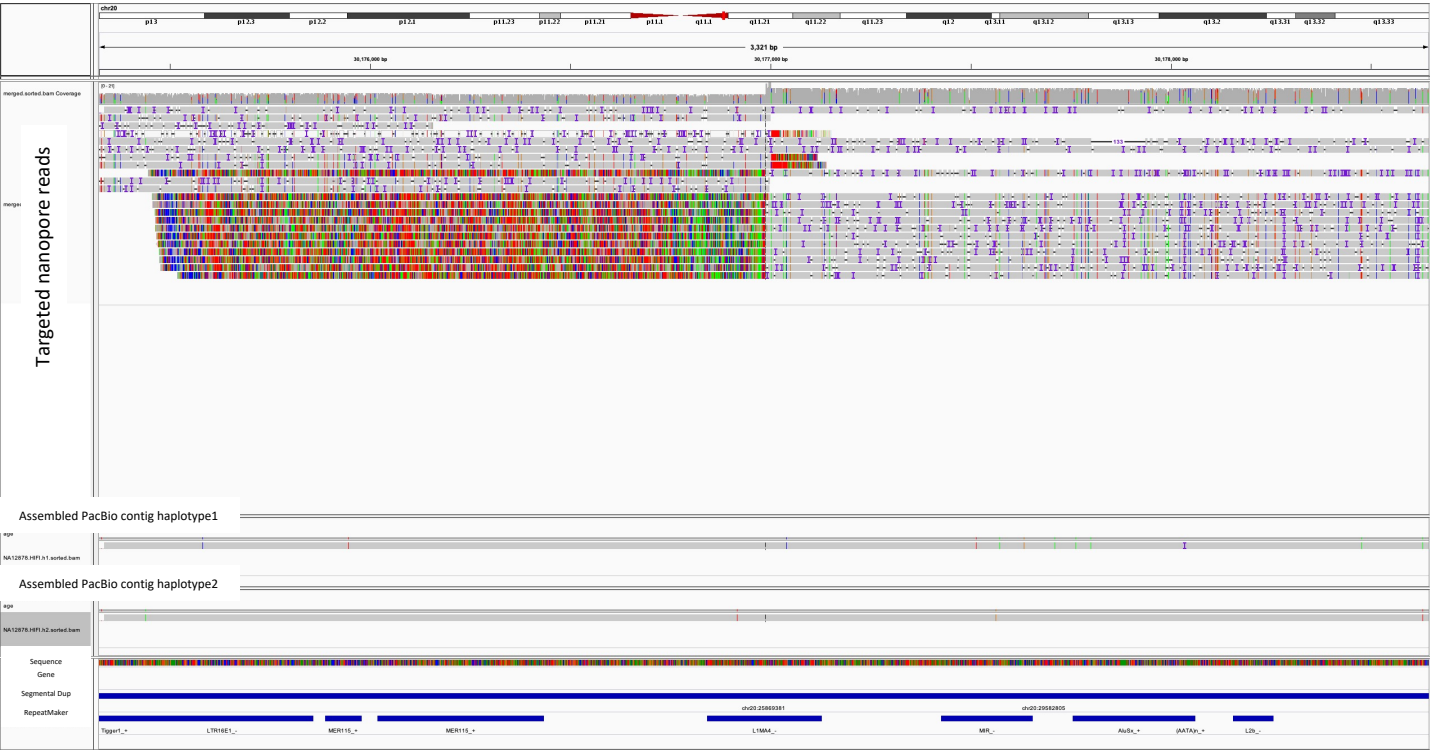
In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 0/32
Allele frequency: 0/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type1 (as missed by other technologies)

chr20:30,175,323-30,178,654



L1Hs call at chrX: 121709076

General information

L1Hs insertion: 403bp L1Hs seq (-) 0/1

5' GACTGGGT TTTTCTTT **Insertion** TTTTCTTT TACTGGTA 3'

Empty site

5' GACTGGGT TTTTCTTT TACTGGTA 3'
3' CTGACCCA AAAAGAAA ATGACCAT 5'

Hallmarks

TSU length: 7bp
polyA length: 75bp
EN Cleavage site: 5' CTTT/A 3'

In Ebert et al. 2021. Science

NA12878 genotype: 0/0
Sample frequency: 0/32
Allele frequency: 0/64

IGV screenshot (general information can be defined by nanopore reads)

True positive type1 (as missed by other technologies)

chrX:121,708,565-121,709,572

