Deletion ~ 58 kb

BCKDHB gene (~239 kb)

Exons: 1 9 10

Deletion ~ 3.7 kb

DBT gene (~63 kb)

Exons: 11

Deletion ~ 19 kb

FH gene (~22 kb)

Exons: 5 1 9

2,318 bp deletion

69 inserted bases

2319 deleted bases

Reference sequence

Patient sequence

2330 bp

69 inserted bases

HPRT1 gene (~40.5 kb)

Exons: 1 5 9

Exon 5

Fa Fb

R