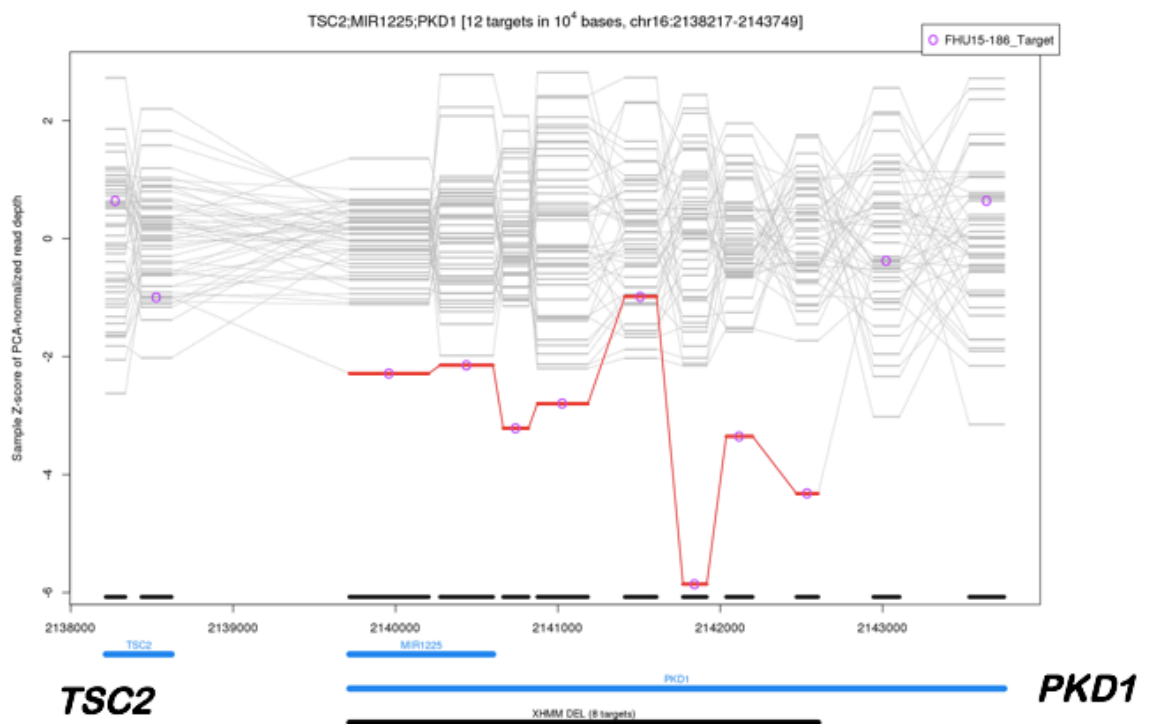
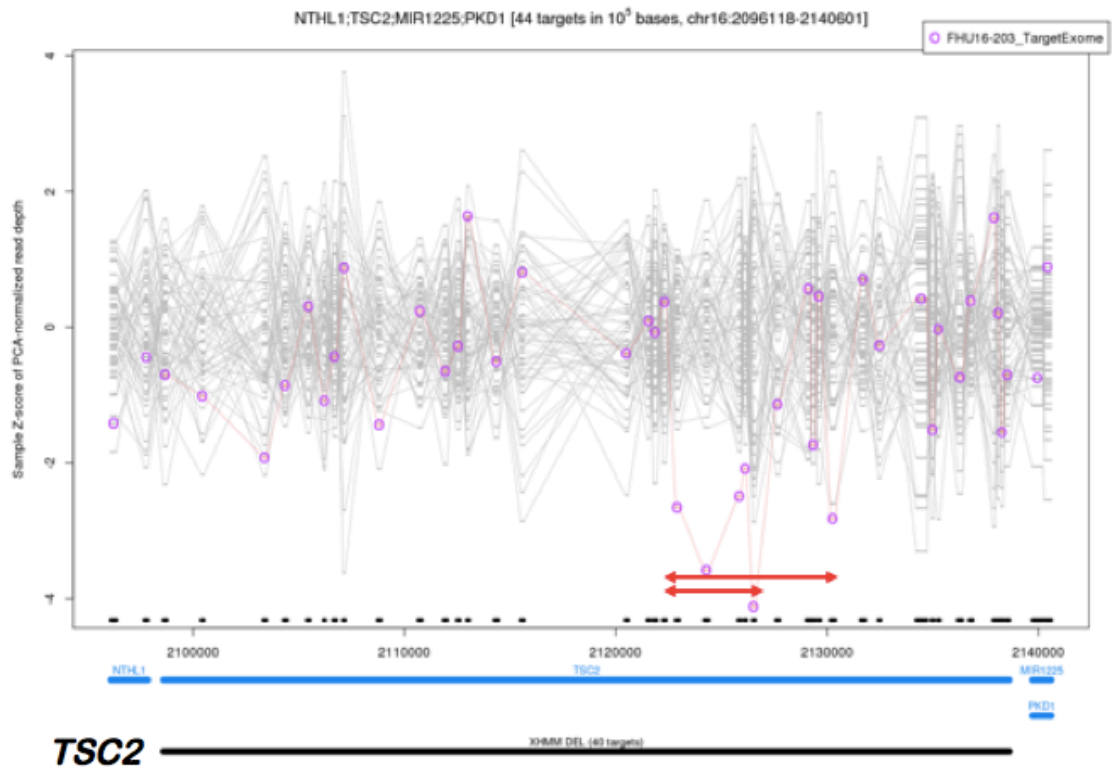
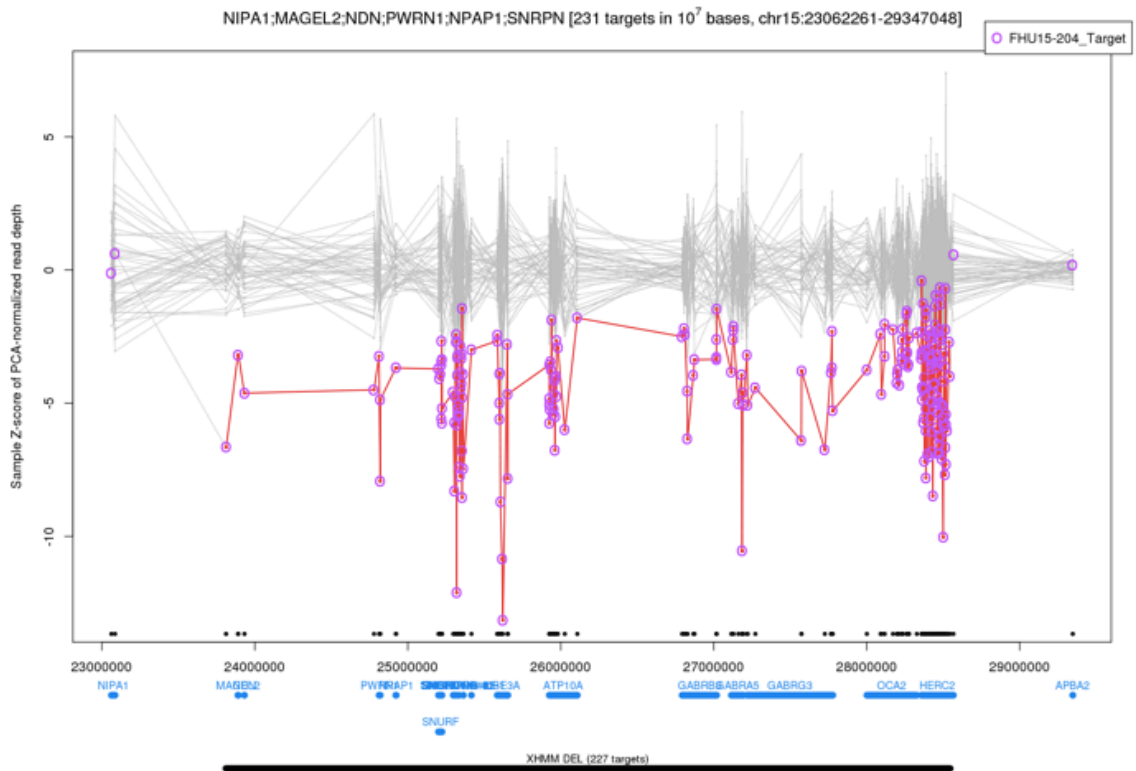


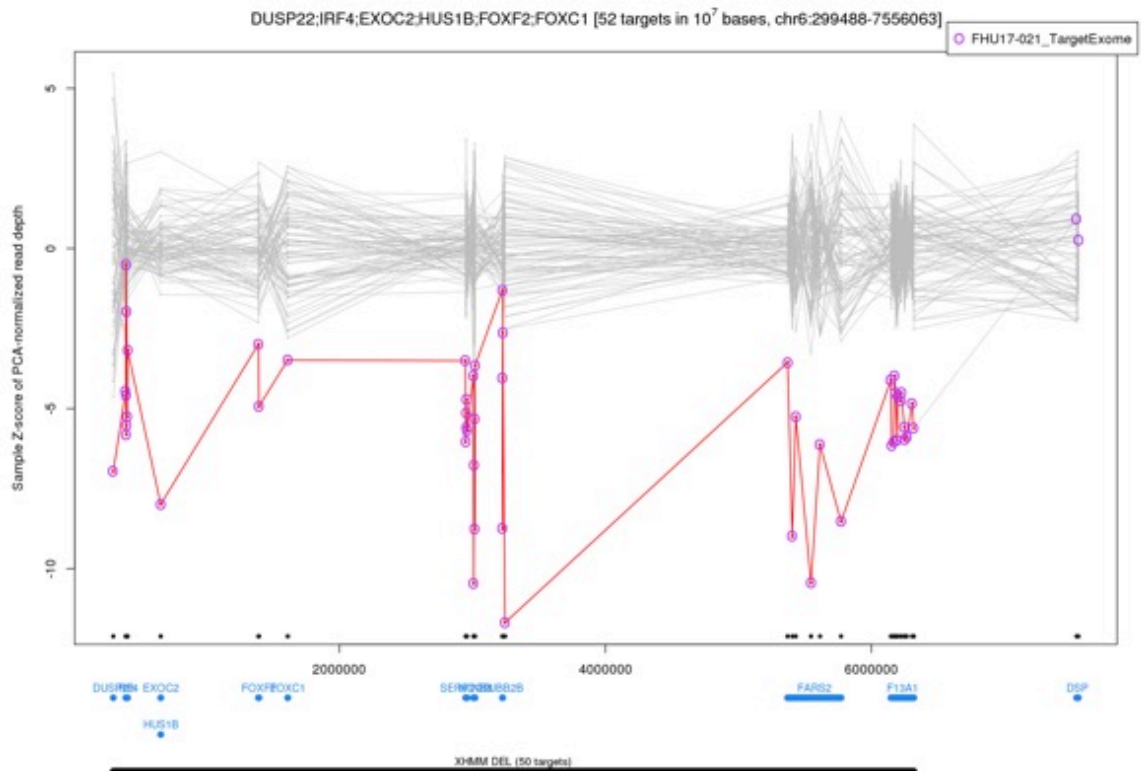
(資料 2)



エクソン単位の欠失・重複



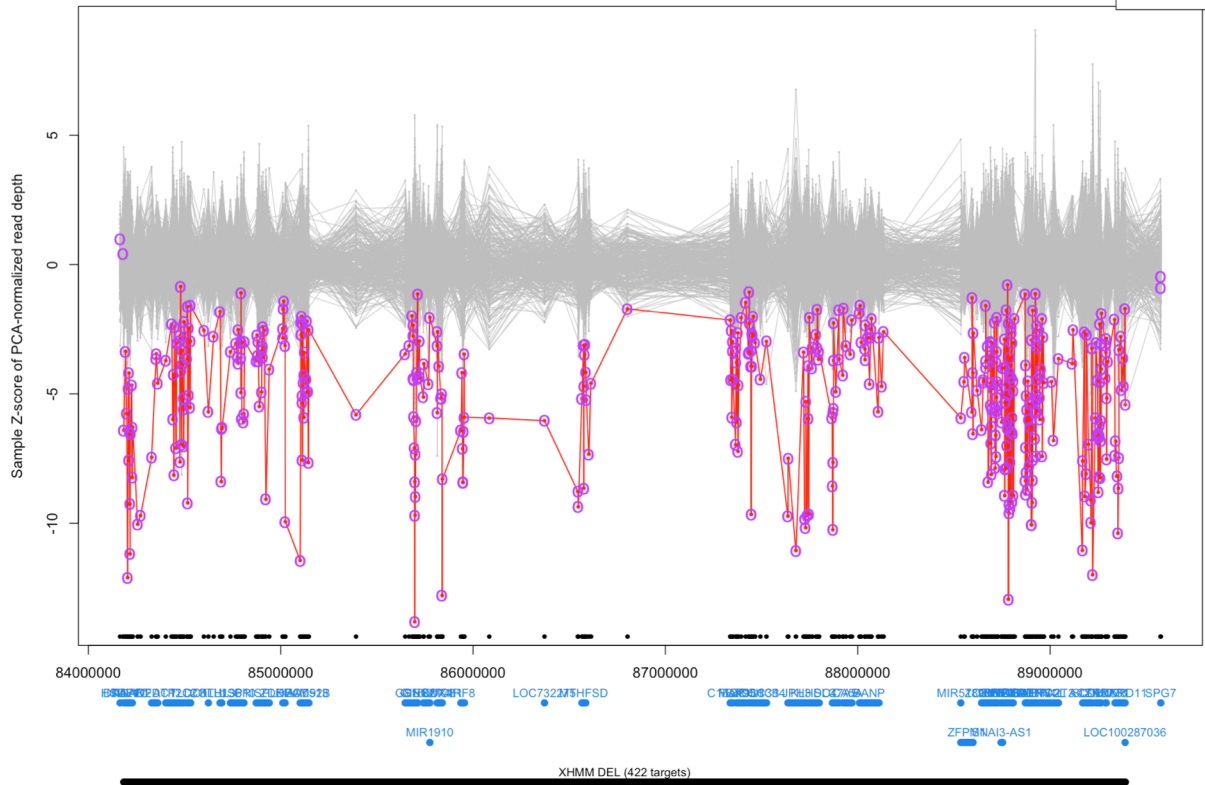
## UBE3A



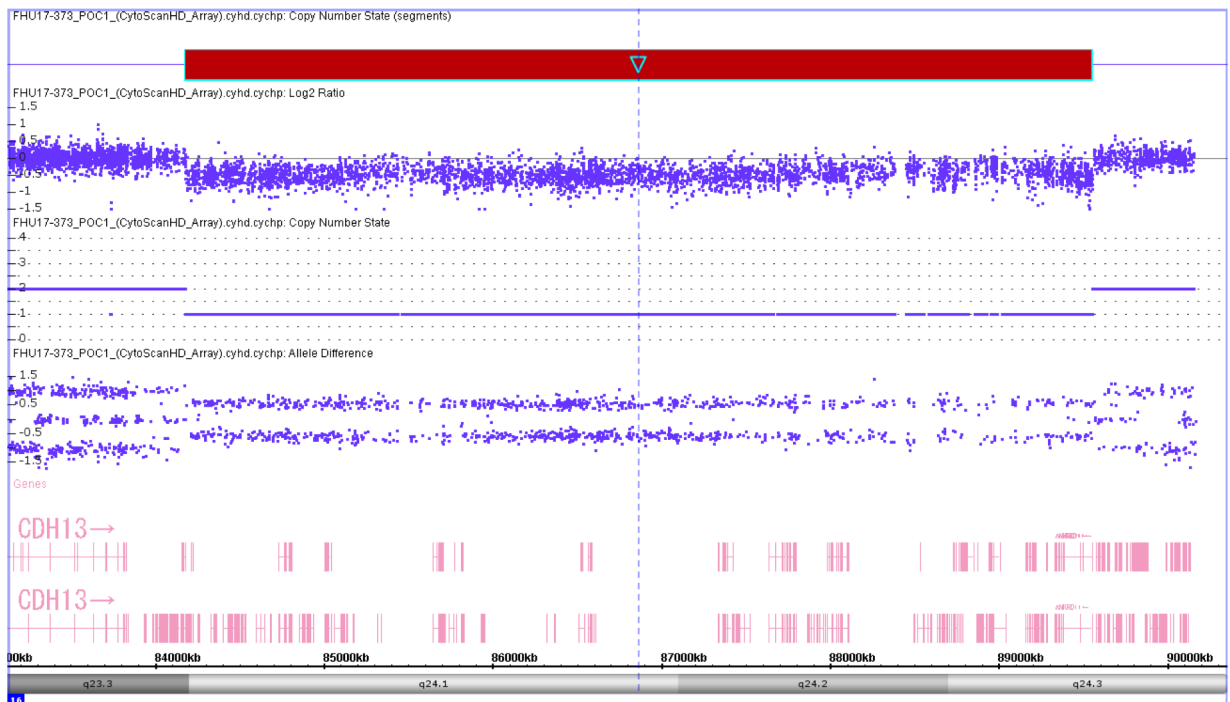
## FOXC1

HSDL1;DNAAF1;TAF1C;ADAD2;WFDC1;ATP2C2 [426 targets in 10<sup>7</sup> bases, chr16:84164556-89575581]

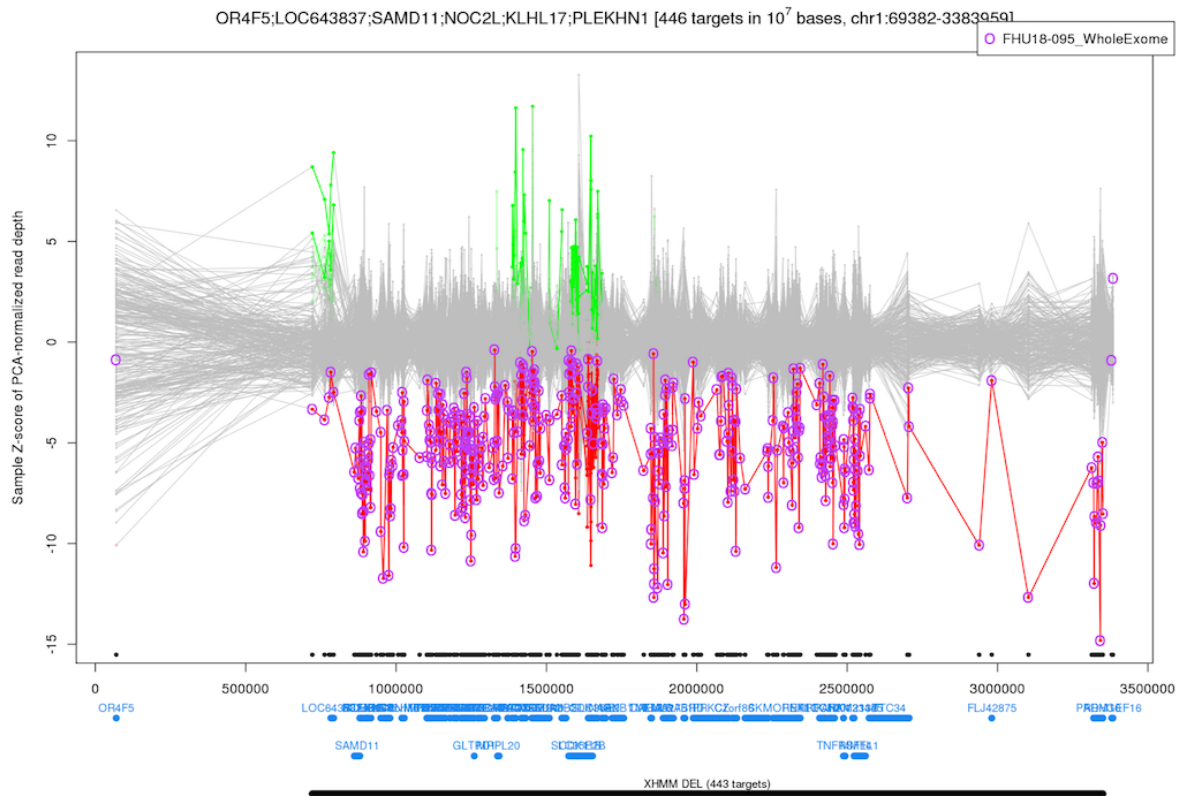
○ FHU17-373



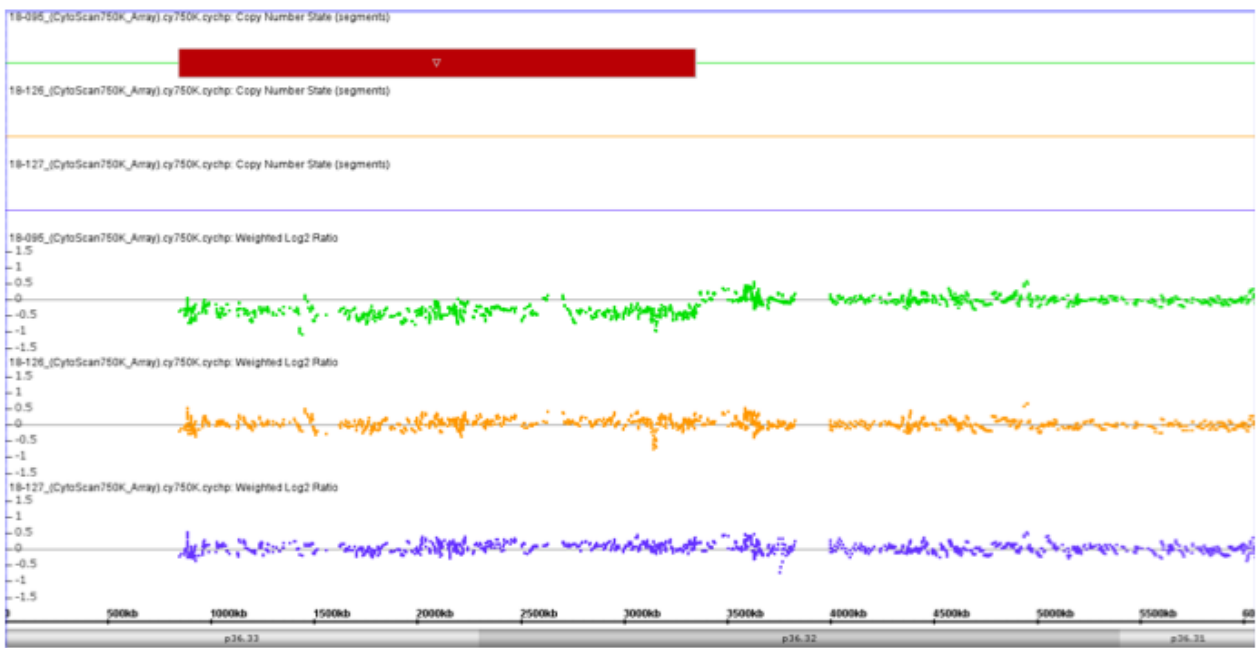
## XHMMによる欠失の同定



## マイクロアレイによる欠失の確認



## XHMMによる欠失の同定



## マイクロアレイによるde novo欠失の確認