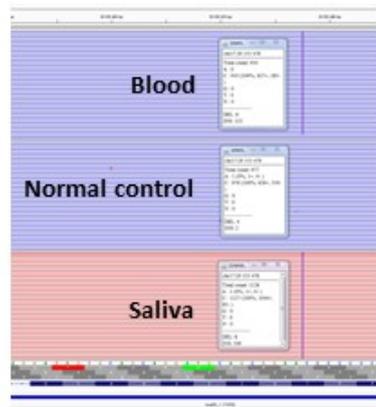


Supplemental Figure 1

Targeted NGS panel (mean depth: 1,500X) identified a mosaic *NF1* pathogenic variant in 3/23 CMMRD patients

IGV screen shots:



Patient 1

NF1 c.2033dup p.(Ile679Aspfs*21)

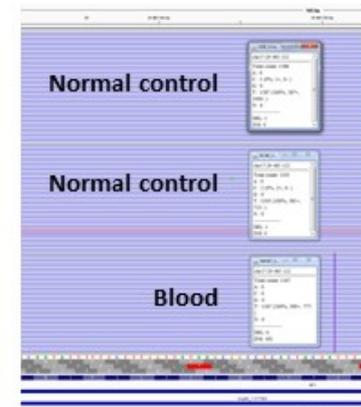
Blood: 153/750 reads = 20%
Saliva: 109/1127 reads = 9%



Patient 2

NF1 c.4751dup p.(Lys1585Glnfs*37)

Blood: reads 100/2560 = 4%



Patient 3

NF1 c.184_185dup p.(Leu62Phefs*2)

Blood: reads 191/1167 : 16%