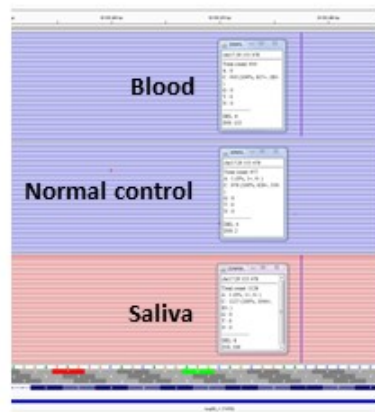


# Supplemental Figure 1

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

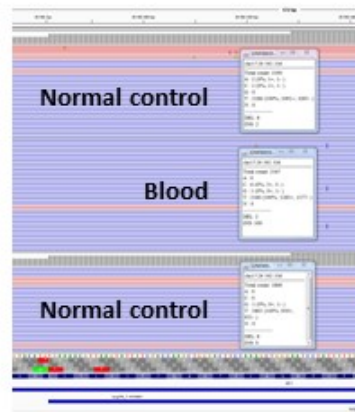
IGV screen shots:



Patient 1

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

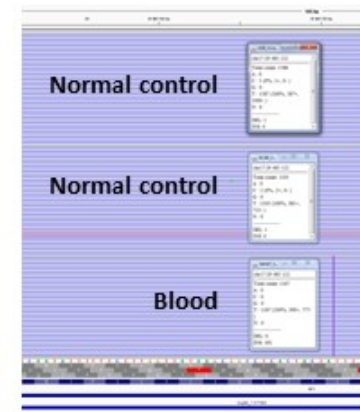
Blood: 153/750 reads = 20%  
Saliva: 109/1127 reads = 9%



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%