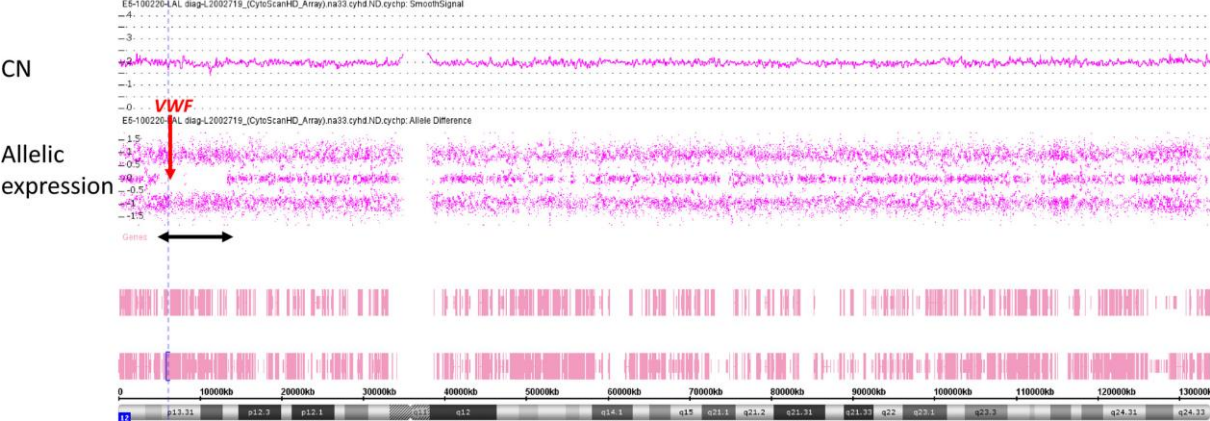


Figure

The homozygous variant p.Gln1311* in exon 28 of VWF is associated with the development of alloantibodies in 3 unrelated patients with type 3 VWD

chr12: 0 - 133,851,894



Tableau

the patients.

regimen inhibitor	ISTH- BAT score	Age at inhibitor detection (years)	Circumstances of detection/number of EDs	Allergic reaction	Known familial consanguinity	Molecular analysis	Anti- VWF:RCo Highest titre, U/ml	Anti- FVIII, U/ml	Anti-VWF (ELISA) Highest dilution positive	Current treatment regimen
s ne age ars rthrosis)	18	18	Lack of therapeutic efficacy/10 years of prophylaxis 2-3 times/week	No	Yes	VWF: p.Gln1311* Hmz	1.5	No	1/1700	High dose prophylaxis with VWF and FVIII plus hormonal therapy
s since of ths osis)	24	1.3	Routine screening/6 EDs	No	Yes	VWF: p.Gln1311* Hmz and F8: c.2113+466_ 2113+473del (intron 13)	15	27	ND	Emicizumab
s since of ths toma)	14	8	Preoperative screening/7 years of prophylaxis 2-3 times/week	No	No	VWF: p.Gln1311* Hmz due to uniparental disomy	0.6	No	1/500	On demand treatment with VWF or rFVIII

days; F, indicates female; Hmz, homozygous; M, male; ND, not determined.