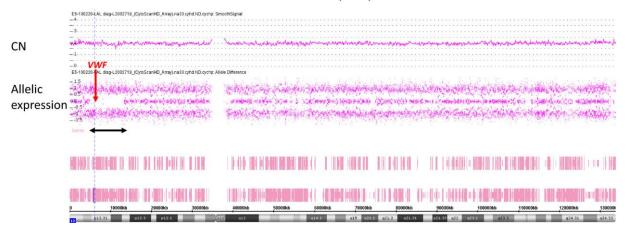
Figure

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





Tableau

the patients.

ISTH- BAT score	Age at inhibitor detection (years)	Circumstances of detection/number of EDs	Allergic reaction	Known familial consanguinity	Mol ecular analysis	Anti- VWF:RCo Highest titre, U/mI	Anti- FVIII, U/ml	Anti-VWF (ELISA) Highest dilution positive	Current treatment regimen
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 F VIII plus hormonal therapy
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
14	8	Preoperative screening/7 years of prophylaxis 2-3 times/week	No	No	VWF: p.Gln1311* Hmz due to uni parental disomy	0.6	No	1/500	On demand treatment with VWF or rFVIII
	BAT score 18	ISTH- BAT detection score (years) 18 18 24 1.3	ISTH- BAT detection (years) 18 18 18 18 18 Lack of therapeutic efficacy/10 years of prophylaxis 2-3 times/week 24 1.3 Routine screening/6 EDs 14 8 Preoperative screening/7 years of prophylaxis 2-3 of prophylaxis 2-3	ISTH- inhibitor detection detection /number score (years) of EDs Allergic reaction 18 18 Lack of therapeutic efficacy/10 years of prophylaxis 2-3 times/week 24 1.3 Routine screening/6 EDs 14 8 Preoperative screening/7 years of prophylaxis 2-3 of prophylaxis 2-3	ISTH- Inhibitor Circumstances of detection / number Allergic Known familial	ISTH- Inhibitor detection Circumstances of detection / number of EDs Allergic Known familial Consanguinity Molecular analysis	ISTH-BAT Inhibitor detection Circumstances of detection /number of EDs Consumption Consumption Circumstances of detection /number of EDs Consumption Consu	ISTH-BAT Inhibitor detection Circumstances of detection /number of EDs Consumption Consump	Age at Inhibitor Inhibitor detection / number detection / number of EDs

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