CNV artéfactuels et MQR sub-optimal

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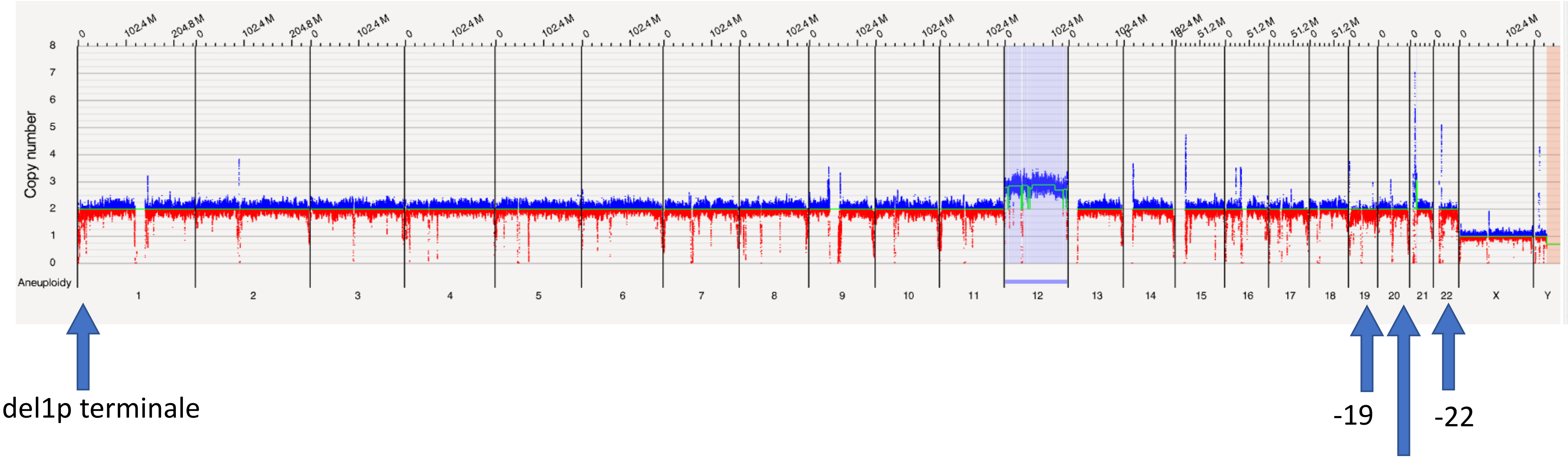
Patients avec MQR suboptimaux

Exemple : patient n°1 : LD=13,57 14-17 NLV=20,1% <15%



del1p terminale

-19

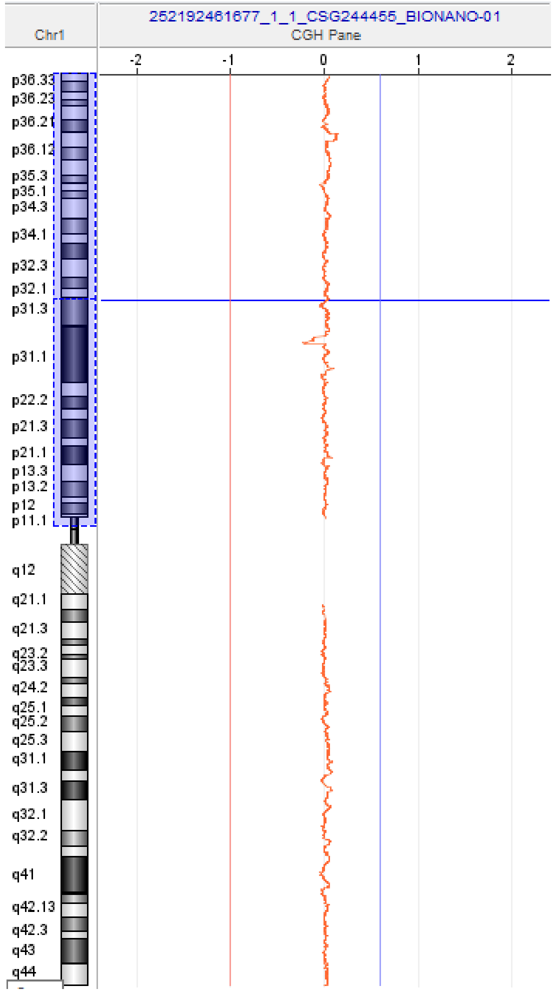
del20q

-22

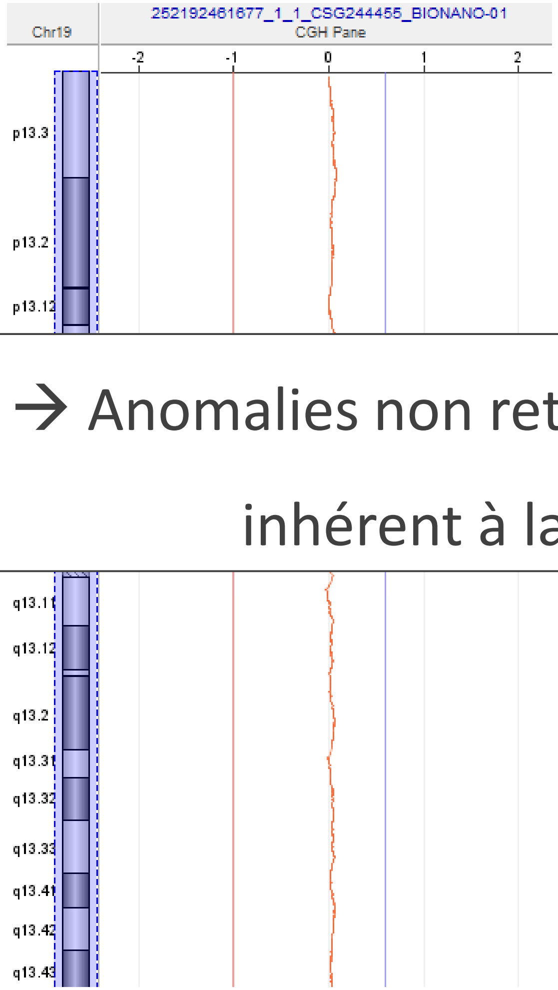
Patients avec MQR suboptimaux

Patient	N50>150kb	N50>20kb	LD	Map rate	PLV	NLV	Profil
	(≥230kb)	*(≥150kb)*	*(14-17/100kb)*	*(>70%)*	*(<10%)*	*(<15%)*	
1	333	273	13,83	75,90%	3,91%	21,19%	del1pter, -19, del20q, -22
2	339	249	14,72	77,60%	4,92%	16,92%	del1pter, -19, del20q, -22
3	314	156	13,98	81%	3,91%	14,86%	del1pter, -19, -22
4	273	198	15,15	77,40%	8%	12,59%	del1pter, -19,-22
5	315	187	15,89	85%	11,62%	12,17%	del1pter, -19, -22
6	307	244	17,12	91%	13,19%	12,00%	-19
7	235	129	14,77	82,80%	4,35%	12,32%	Ø
8	360	304	17,59	90,30%	11,46%	10,81%	Ø
9	290	216	14,27	79%	10,35%	13,10%	Ø

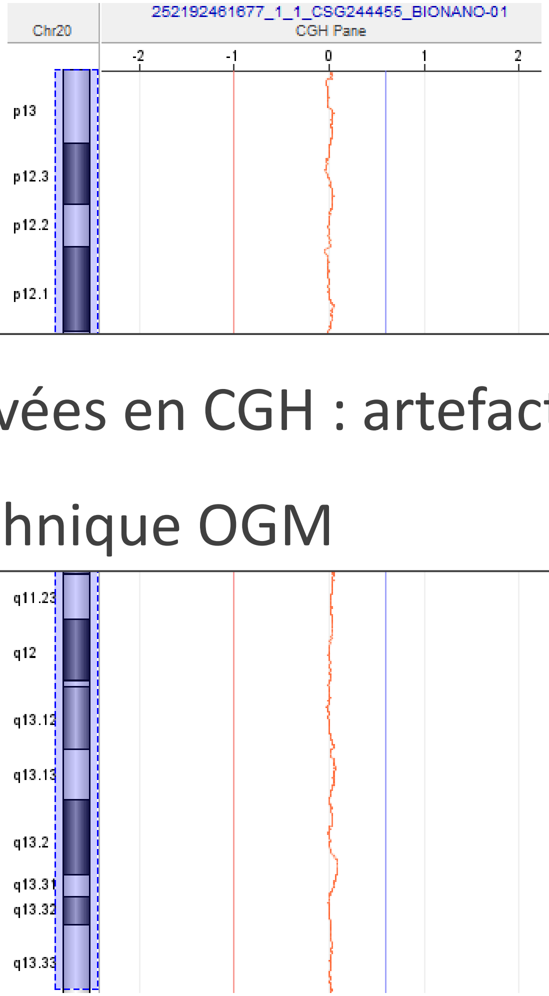
Résultats CGH :



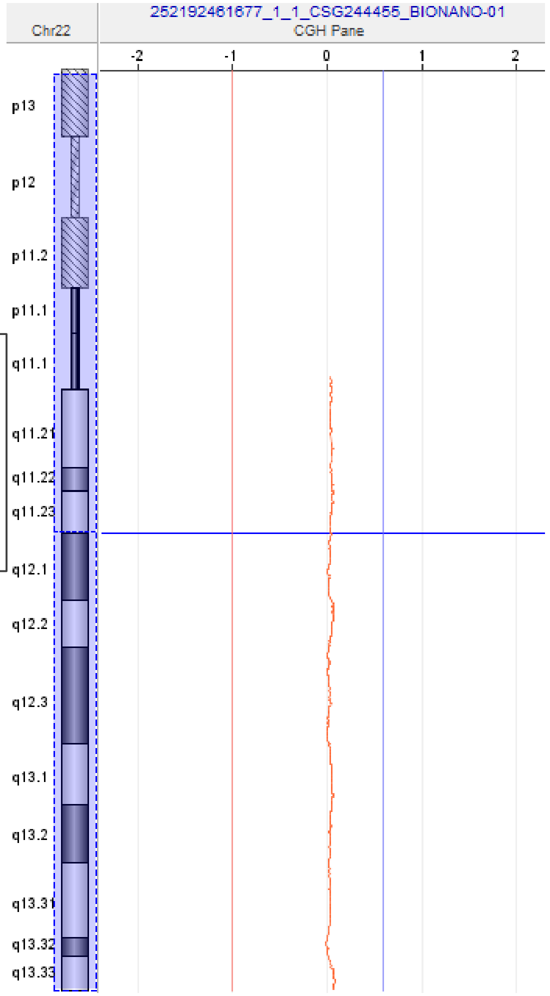
Chr 1



Chr 19



Chr 20

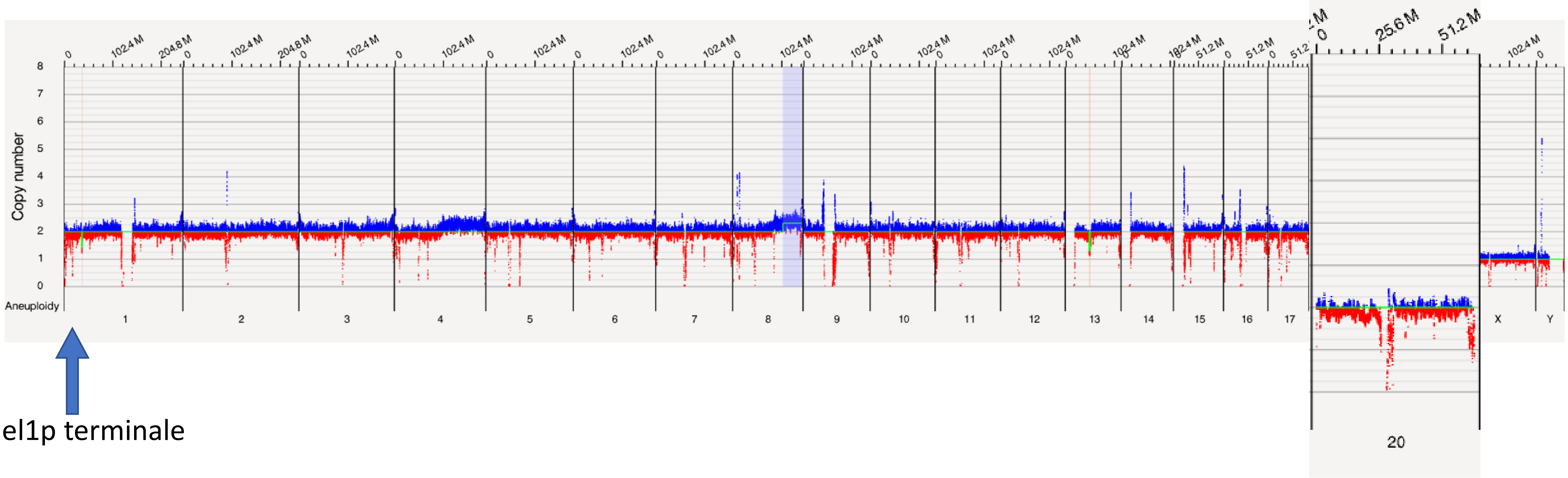


Chr 22

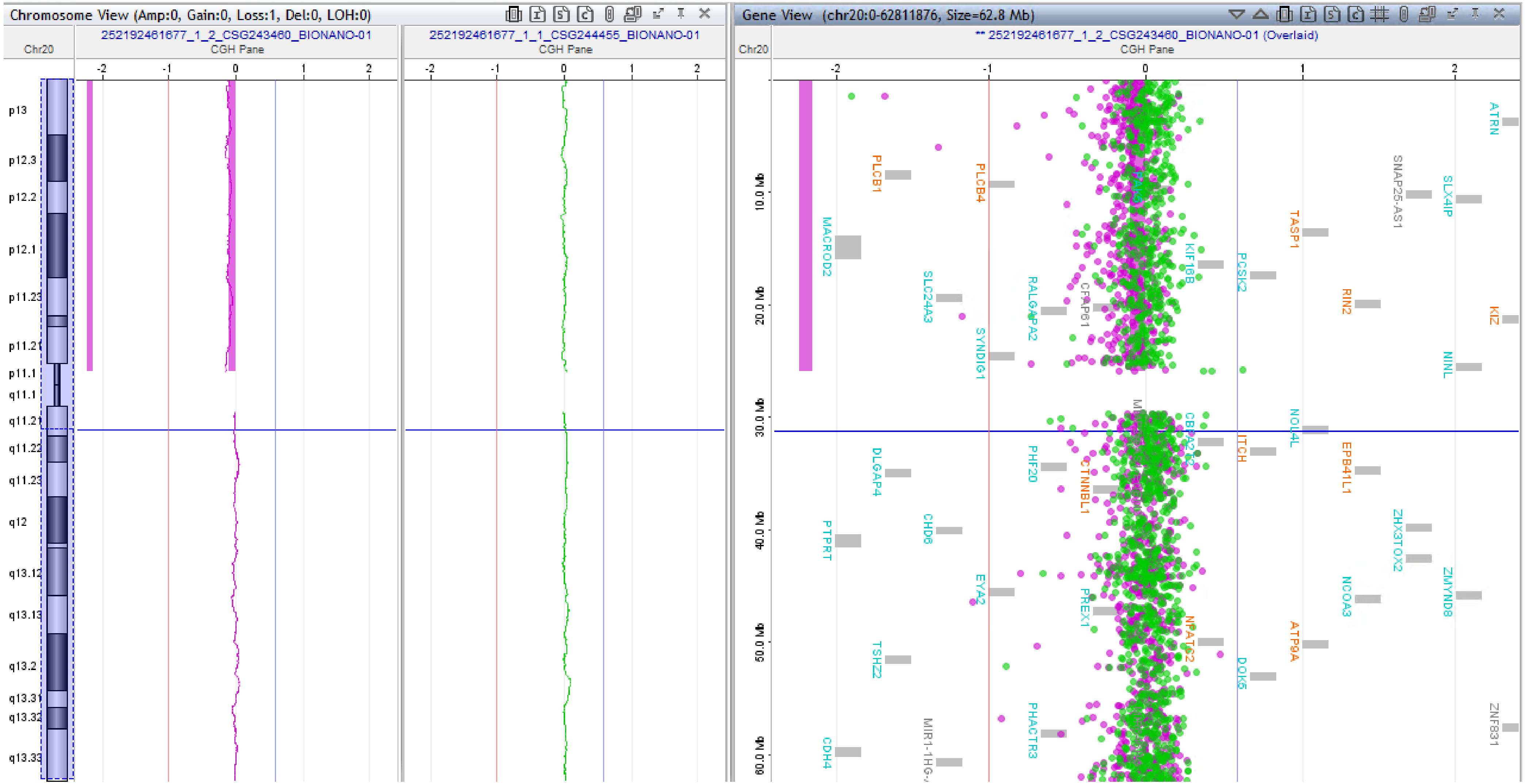
→ Anomalies non retrouvées en CGH : artefact inhérent à la technique OGM

Patient n°2 : NLV=16,5%   <15%

Monosomie 20 ?



del1p terminale



Patient n°4 avec del20p

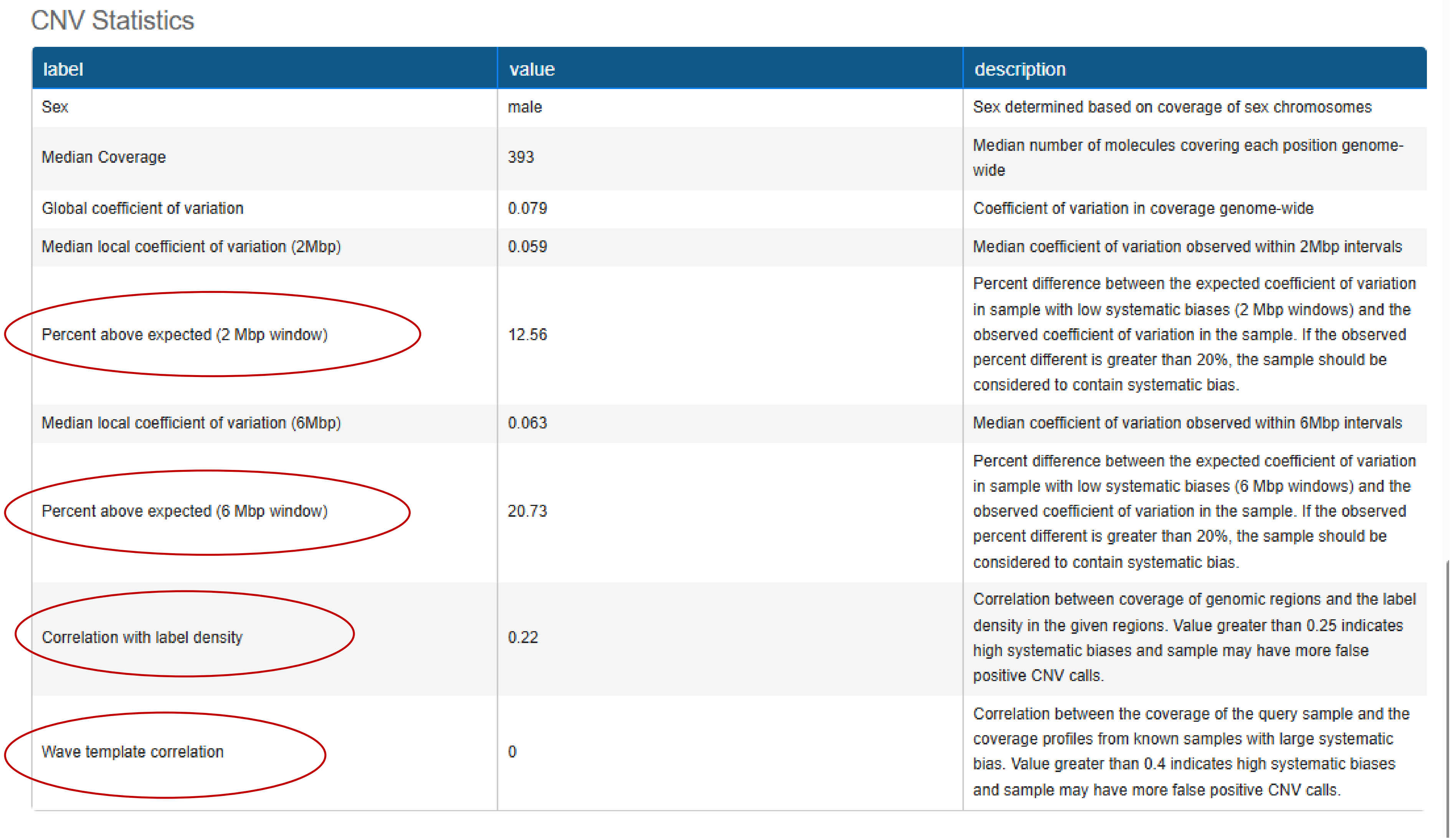
Patient sans anomalie

- CGH : del20p présente mais pas de del 20q
- OGM : aspect de monosomie 20 qui masque la vraie del20p

Patient	N50>150kb	N50>20kb	LD	Map rate	PLV	NLV	Profil
	(≥230kb)	(≥150kb)	(14-17/100kb)	(>70%)	(<10%)	(<15%)	
1	333	273	13,83	75,90%	3,91%	21,19%	del1pter, -19, del20q, -22
2	339	249	14,72	77,60%	4,92%	16,92%	del1pter, -19, del20q, -22
3	314	156	13,98	81%	3,91%	14,86%	del1pter, -19, -22
4	273	198	15,15	77,40%	8%	12,59%	del1pter, -19,-22
5	315	187	15,89	85%	11,62%	12,17%	del1pter, -19, -22
6	307	244	17,12	91%	13,19%	12,00%	-19
7	235	129	14,77	82,80%	4,35%	12,32%	Ø
8	360	304	17,59	90,30%	11,46%	10,81%	Ø
9	290	216	14,27	79%	10,35%	13,10%	Ø

NLV : Percentage of reference labels absent in molecules
PLV : Percentage of labels absent in reference

Informatic Report :



CNV Statistics

label	value	description
Sex	male	Sex determined based on coverage of sex chromosomes
Median Coverage	393	Median number of molecules covering each position genome-wide
Global coefficient of variation	0.079	Coefficient of variation in coverage genome-wide
Median local coefficient of variation (2Mbp)	0.059	Median coefficient of variation observed within 2Mbp intervals
Percent above expected (2 Mbp window)	12.56	Percent difference between the expected coefficient of variation in sample with low systematic biases (2 Mbp windows) and the observed coefficient of variation in the sample. If the observed percent different is greater than 20%, the sample should be considered to contain systematic bias.
Median local coefficient of variation (6Mbp)	0.063	Median coefficient of variation observed within 6Mbp intervals
Percent above expected (6 Mbp window)	20.73	Percent difference between the expected coefficient of variation in sample with low systematic biases (6 Mbp windows) and the observed coefficient of variation in the sample. If the observed percent different is greater than 20%, the sample should be considered to contain systematic bias.
Correlation with label density	0.22	Correlation between coverage of genomic regions and the label density in the given regions. Value greater than 0.25 indicates high systematic biases and sample may have more false positive CNV calls.
Wave template correlation	0	Correlation between the coverage of the query sample and the coverage profiles from known samples with large systematic bias. Value greater than 0.4 indicates high systematic biases and sample may have more false positive CNV calls.

Patient	N50>150kb	N50>20kb	LD	Map rate	PLV	NLV	Profil	% above expected (2Mbp window) (<	% above expected (6Mbp window) (<	correlation with label density (< 0,25)
	(≥230kb)	*(≥150kb)*	*(14-17/100kb)*	*(>70%)*	*(<10%)*	*(<15%)*				
1	333	273	13,83	75,90%	3,91%	21,19%	del1pter, -19, del20q, -22	12,56	20,73	0,22
2	339	249	14,72	77,60%	4,92%	16,92%	del1pter, -19, del20q, -22	0,11	12,7	0,22
3	314	156	13,98	81%	3,91%	14,86%	del1pter, -19, -22	6,15	14,37	0,3
4	273	198	15,15	77,40%	8%	12,59%	del1pter, -19,-22	7,86	13,95	0,27
5	315	187	15,89	85%	11,62%	12,17%	del1pter, -19, -22	1,67	11,76	0,18
6	307	244	17,12	91%	13,19%	12,00%	-19	0,87	7,86	0,16
7	235	129	14,77	82,80%	4,35%	12,32%	Ø	6,65	14,51	0,09
8	360	304	17,59	90,30%	11,46%	10,81%	Ø	-1,73	8,56	0,14
9	290	216	14,27	79%	10,35%	13,10%	Ø	-1,64	6,32	0,091

NLV : Percentage of reference labels absent in molecules
PLV : Percentage of labels absent in reference

	% above expected (2Mbp window) (< 20%)	% above expected (6Mbp window) (< 20%)	correlation with label density (< 0,25)
moy	-4,94	3,80	0,0937
sd	3,60	3,56	0,0366
moy+3ds	**5,87**	**14,50**	**0,2036**

Conclusion

MQR sub-optimaux :

1- CNV artéfactuels de type del1pter, -19, del20q, -22

2- on ne met pas en évidence un marqueur prépondérant :
vérifier sur l'informatic report les « metrics » CNV