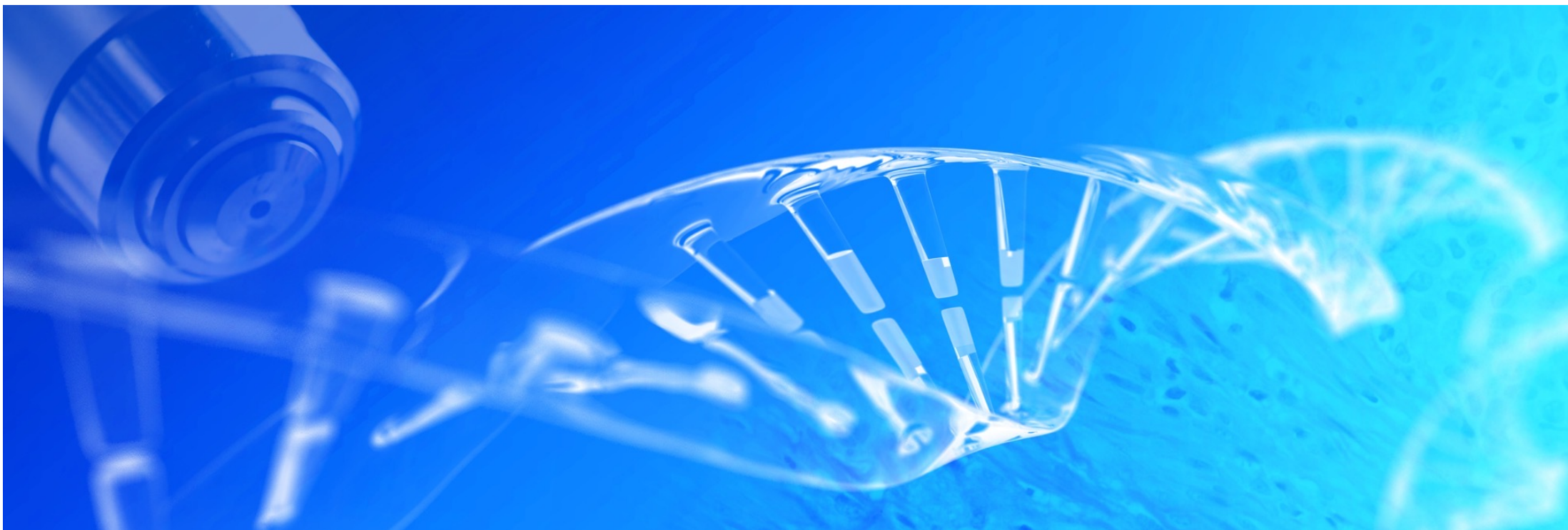




Programmes nationaux d'EEQ des examens moléculaires

Réunion de restitution de la campagne 2024 – 18 mars 2025

Programme NGS



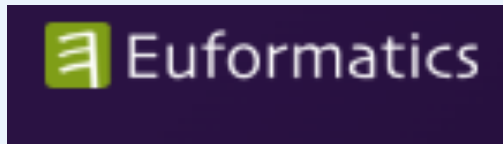
Programme Analyse des données bio-informatiques

Echantillons

- 24-MULTI-10
- 24-BRCA-06

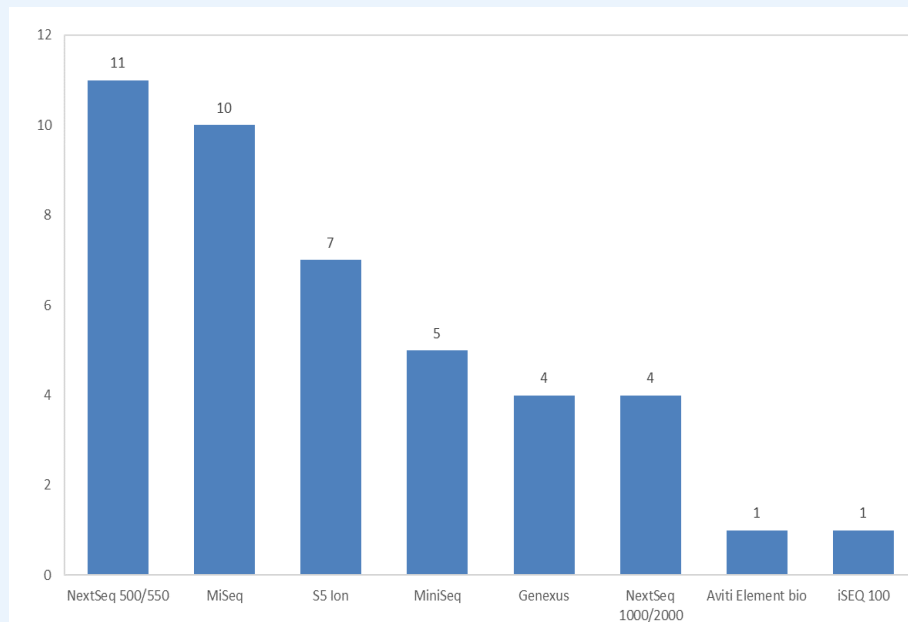
Fichiers

- BAM
- VCF
- BED interne ou BED générique

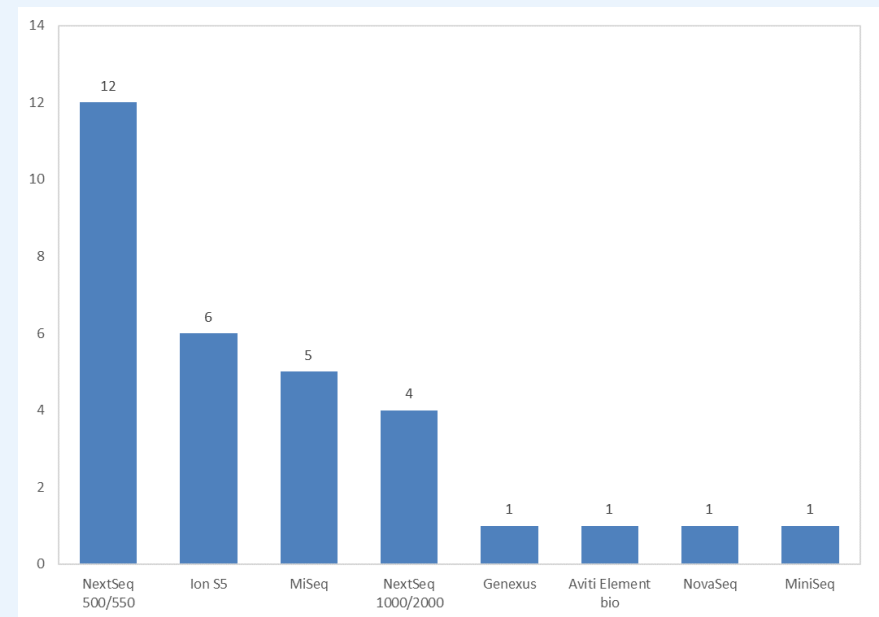


Statistiques

Multiparamétrique: 43 participants



Ovaire: 31 participants



Critères d'évaluation

Critères de qualité

- Nombre de reads totaux
- Uniformité
- Base quality
- Off target
- Read depth / mapped
- Coverage 1X et 100X
- Ti/Tv
- Nombre de SNP et Indel

Critères d'évaluation

BED

- Bed Gen&Tiss
- Bed proposé par le participant
- Bed minimal – 6 gènes / BRCA1+BRCA2
- Bed commun
- Profondeurs – locus BRCA1 et PIK3CA

BED Commun

Bed 6 gènes

NRAS	Exon4	BRAF	Exon18	KIT	Exon8	EGFR	Exon10
NRAS	Exon3	BRAF	Exon17	KIT	Exon9	EGFR	Exon11
NRAS	Exon2	BRAF	Exon16	KIT	Exon10	EGFR	Exon12
PIK3CA	Exon2	BRAF	Exon15	KIT	Exon11	EGFR	Exon13
PIK3CA	Exon3	BRAF	Exon14	KIT	Exon12	EGFR	Exon14
PIK3CA	Exon4	BRAF	Exon13	KIT	Exon13	EGFR	Exon15
PIK3CA	Exon5	BRAF	Exon12	KIT	Exon14	EGFR	Exon16
PIK3CA	Exon6	BRAF	Exon11	KIT	Exon15	EGFR	Exon17
PIK3CA	Exon7	BRAF	Exon10	KIT	Exon16	EGFR	Exon18
PIK3CA	Exon8	BRAF	Exon9	KIT	Exon17	EGFR	Exon19
PIK3CA	Exon9	BRAF	Exon8	KIT	Exon18	EGFR	Exon20
PIK3CA	Exon10	BRAF	Exon7	KIT	Exon19	EGFR	Exon21
PIK3CA	Exon11	BRAF	Exon6	KIT	Exon20	EGFR	Exon22
PIK3CA	Exon12	BRAF	Exon5	KIT	Exon21	EGFR	Exon23
PIK3CA	Exon13	BRAF	Exon4	EGFR	Exon2	EGFR	Exon24
PIK3CA	Exon14	BRAF	Exon3	EGFR	Exon3	EGFR	Exon25
PIK3CA	Exon15	BRAF	Exon2	EGFR	Exon4	EGFR	Exon26
PIK3CA	Exon16	BRAF	Exon1	EGFR	Exon5		
PIK3CA	Exon17	KRAS	Exon5	EGFR	Exon6		
PIK3CA	Exon18	KRAS	Exon4	EGFR	Exon7		
PIK3CA	Exon19	KRAS	Exon3	EGFR	Exon8		
PIK3CA	Exon20	KRAS	Exon2	EGFR	Exon9		
PIK3CA	Exon21						

Bed BRCA1/BRCA2

BRCA2	Exon8	BRCA1	Exon24
BRCA2	Exon9	BRCA1	Exon24
BRCA2	Exon10	BRCA1	Exon23
BRCA2	Exon11	BRCA1	Exon22
BRCA2	Exon12	BRCA1	Exon21
BRCA2	Exon13	BRCA1	Exon20
BRCA2	Exon14	BRCA1	Exon19
BRCA2	Exon15	BRCA1	Exon18
BRCA2	Exon16	BRCA1	Exon17
BRCA2	Exon17	BRCA1	Exon16
BRCA2	Exon18	BRCA1	Exon15
BRCA2	Exon19	BRCA1	Exon14
BRCA2	Exon20	BRCA1	Exon13
BRCA2	Exon21	BRCA1	Exon12
BRCA2	Exon22	BRCA1	Exon11
BRCA2	Exon23/24	BRCA1	Exon10
BRCA2	Exon25	BRCA1	Exon9
BRCA2	Exon26	BRCA1	Exon8
BRCA2	Exon27	BRCA1	Exon7
BRCA2	Exon27+3'UT	BRCA1	Exon6
		BRCA1	Exon5
		BRCA1	Exon3
		BRCA1	Exon2

Evaluation

Déterminer la variabilité

Identifier des extrêmes

Critères – Multiparamétrique

	Moyenne de Reads total
NextSeq 1000/2000	11 336 249
NextSeq 500/550	6 142 513
Aviti Element bio	3 294 692
MiniSeq	2 979 965
Genexus	1 511 558
MiSeq	1 504 432
iSEQ 100	855 983
S5 lon	736 882

	Moyenne de Read depth (ave)
iSEQ 100	5 173
MiniSeq	3 966
NextSeq 500/550	3 283
Genexus	2 783
S5 lon	1 964
MiSeq	1 531
NextSeq 1000/2000	1 489
Aviti Element bio	1 239

	Moyenne de Base quality (average)
Aviti Element bio	84
MiSeq	40
NextSeq 500/550	37
NextSeq 1000/2000	36
MiniSeq	35
S5 lon	28
iSEQ 100	28
Genexus	25

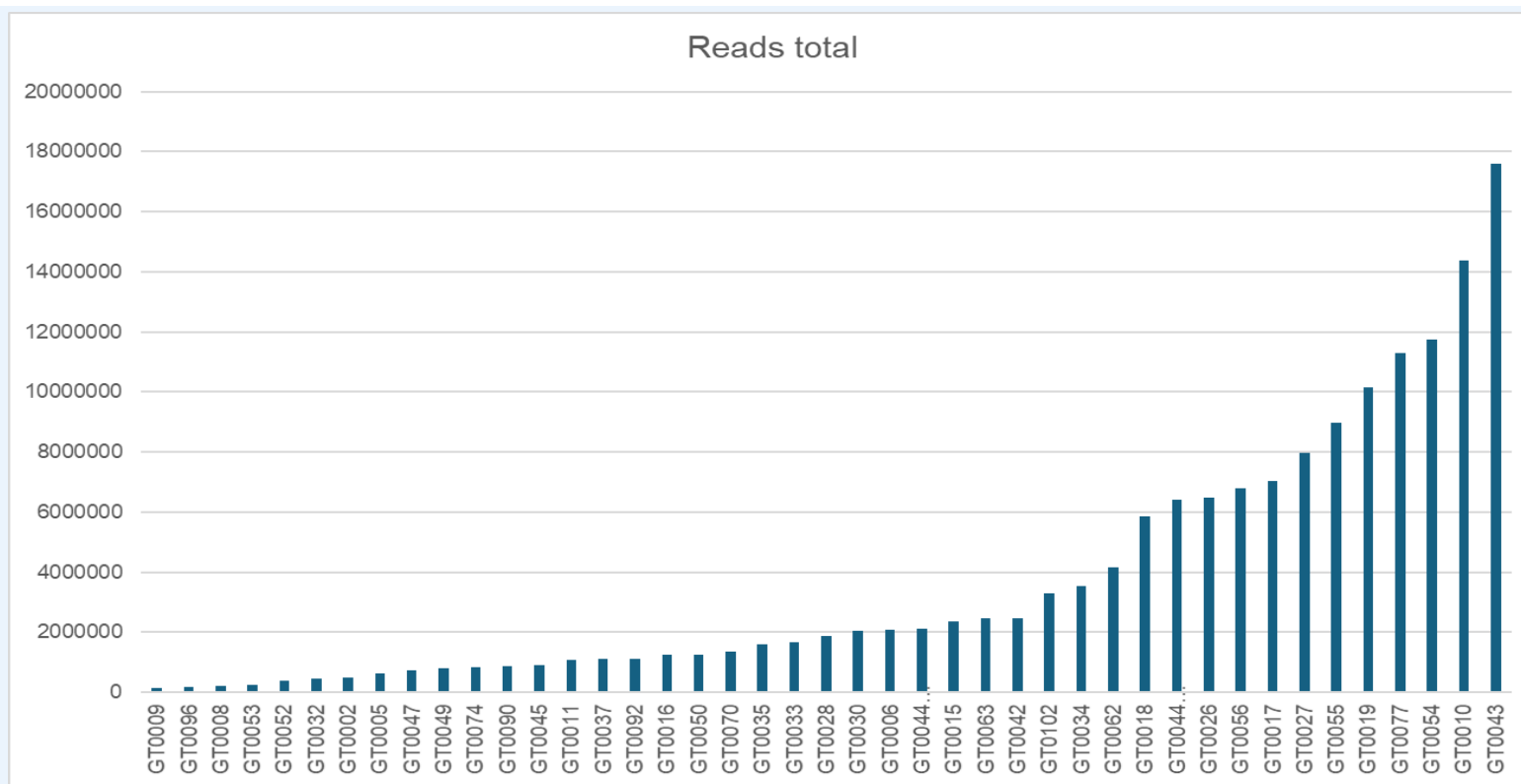
	Moyenne de Coverage at 1X (%)
Aviti Element bio	100
NextSeq 1000/2000	92
MiniSeq	91
NextSeq 500/550	85
MiSeq	78
iSEQ 100	75
Genexus	57
S5 lon	56

	Moyenne de Uniformity (%)
Aviti Element bio	100
MiniSeq	89
NextSeq 1000/2000	87
NextSeq 500/550	79
iSEQ 100	72
MiSeq	67
Genexus	56
S5 lon	47

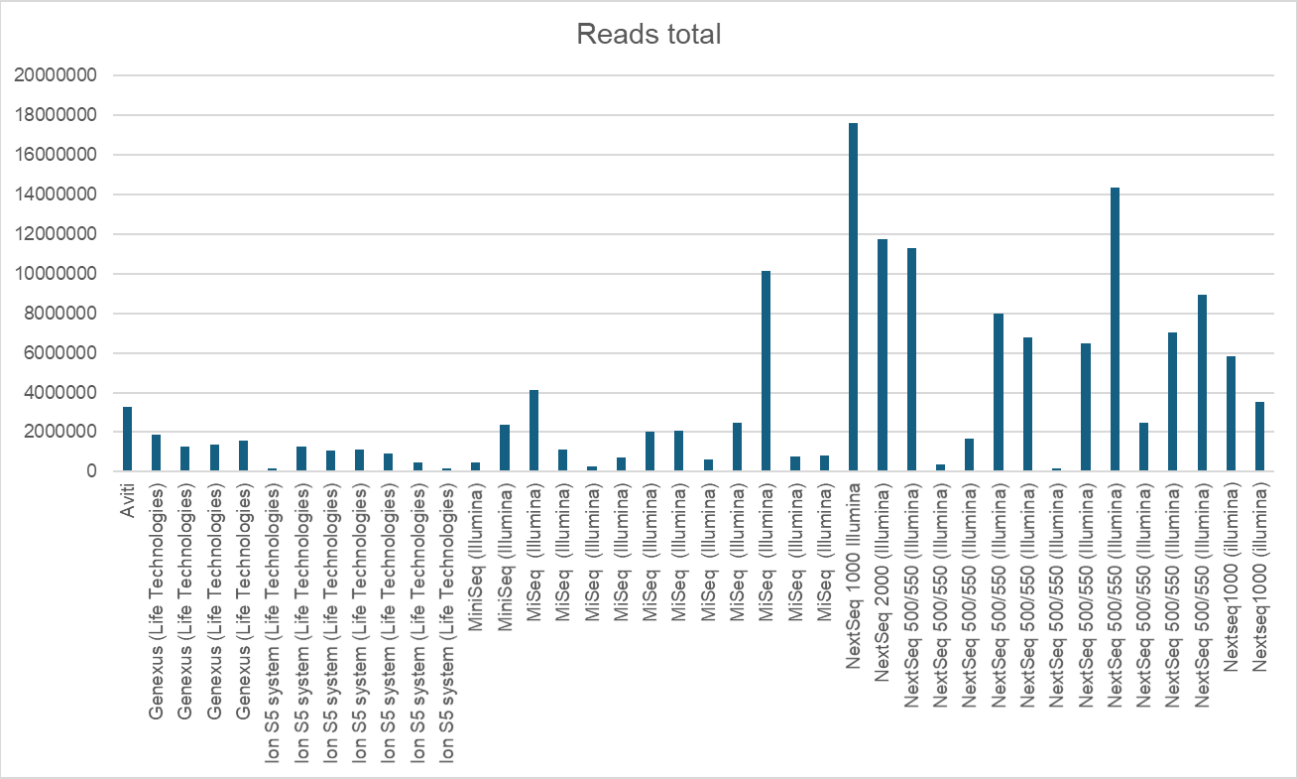
	Moyenne de Coverage at 100X (%)
Aviti Element bio	100
NextSeq 1000/2000	88
MiniSeq	81
NextSeq 500/550	78
MiSeq	75
iSEQ 100	72
Genexus	56
S5 lon	49

	Moyenne de Ti/Tv
Aviti Element bio	15
MiniSeq	6
MiSeq	3
S5 lon	3
NextSeq 1000/2000	3
NextSeq 500/550	2
iSEQ 100	2
Genexus	1

Multiparamétrique - Nombre de reads totaux

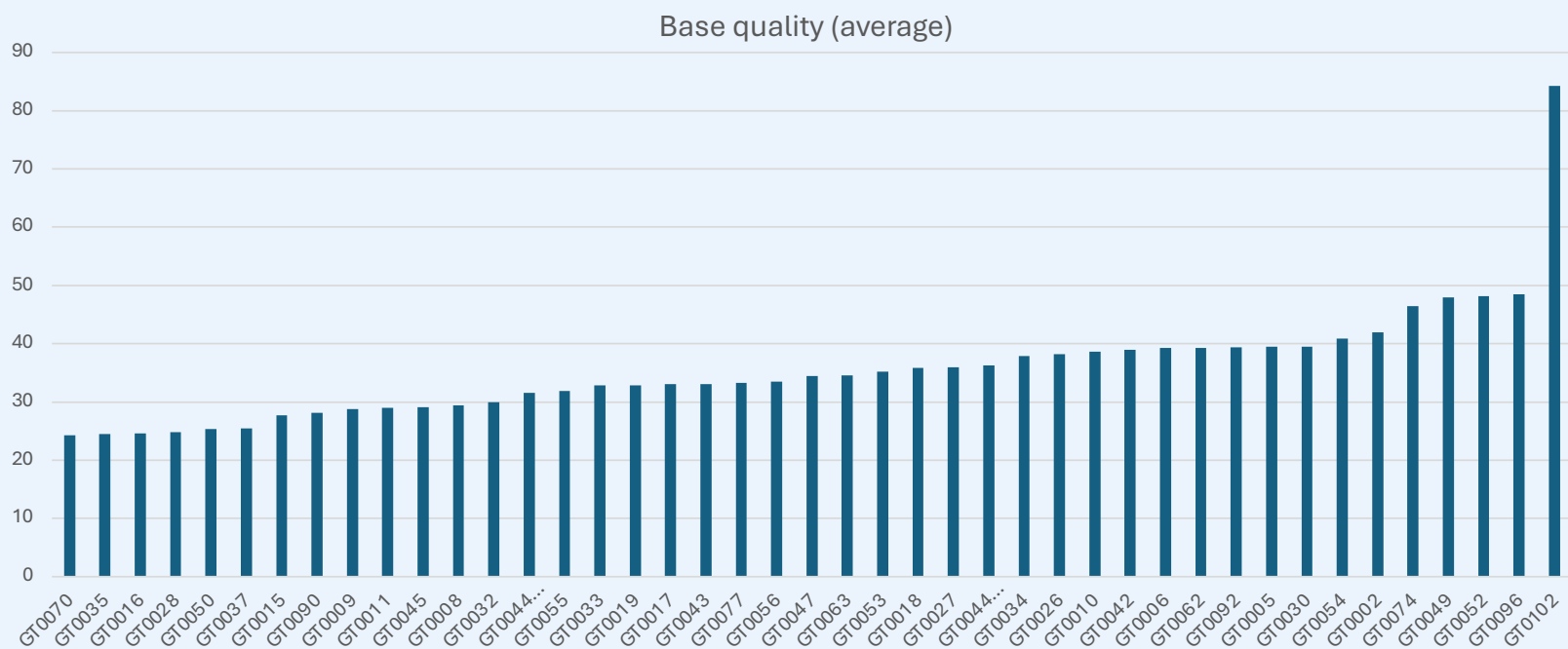


Multiparamétrique - Nombre de reads totaux

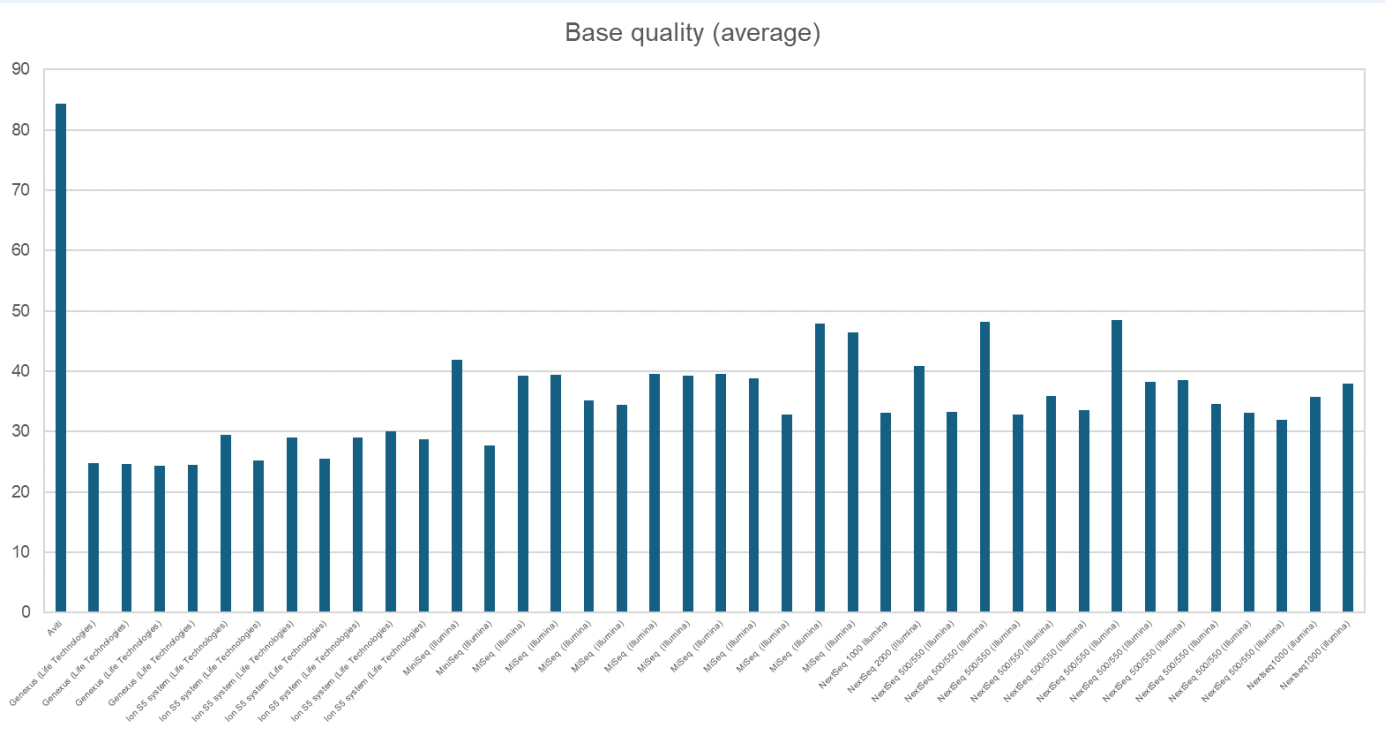


	Moyenne de Reads total
NextSeq 1000/2000	11 336 249
NextSeq 500/550	6 142 513
Aviti Element bio	3 294 692
MiniSeq	2 979 965
Genexus	1 511 558
MiSeq	1 504 432
iSEQ 100	855 983
S5 Ion	736 882

Multiparamétrique – Base quality

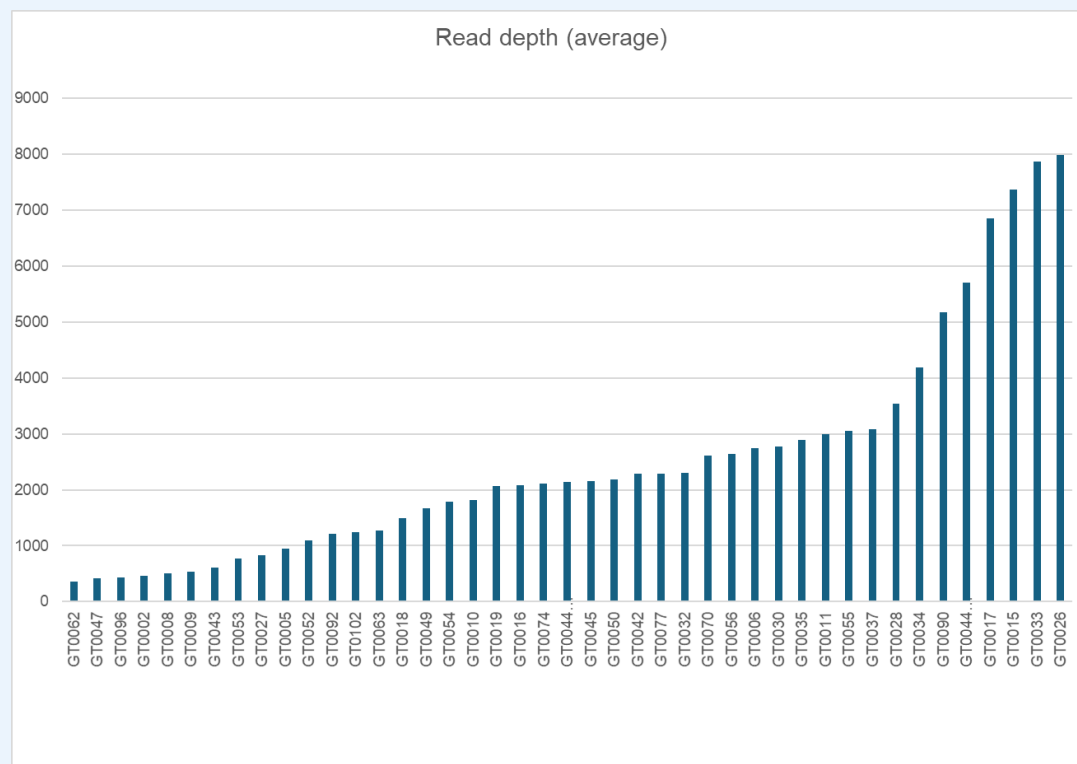


Multiparamétrique – Base quality



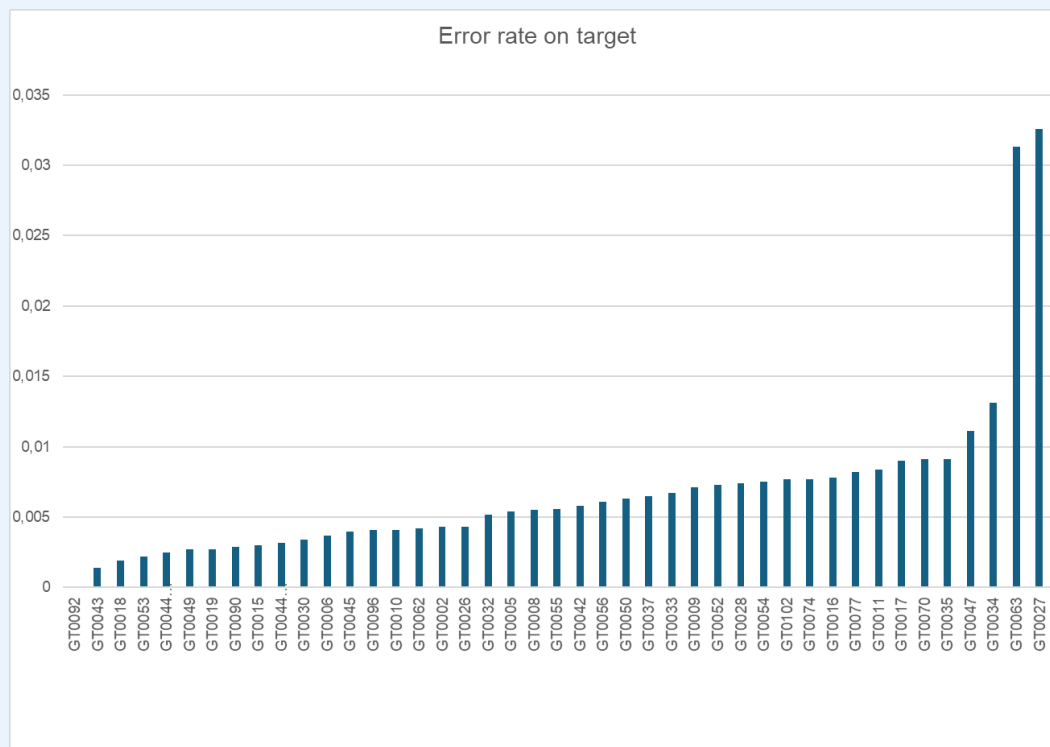
	Moyenne de Base quality (average)
Aviti Element bio	84
MiSeq	40
NextSeq 500/550	37
NextSeq 1000/2000	36
MiniSeq	35
S5 Ion	28
iSEQ 100	28
Genexus	25

Multiparamétrique – Read depth, Bed à 6 gènes



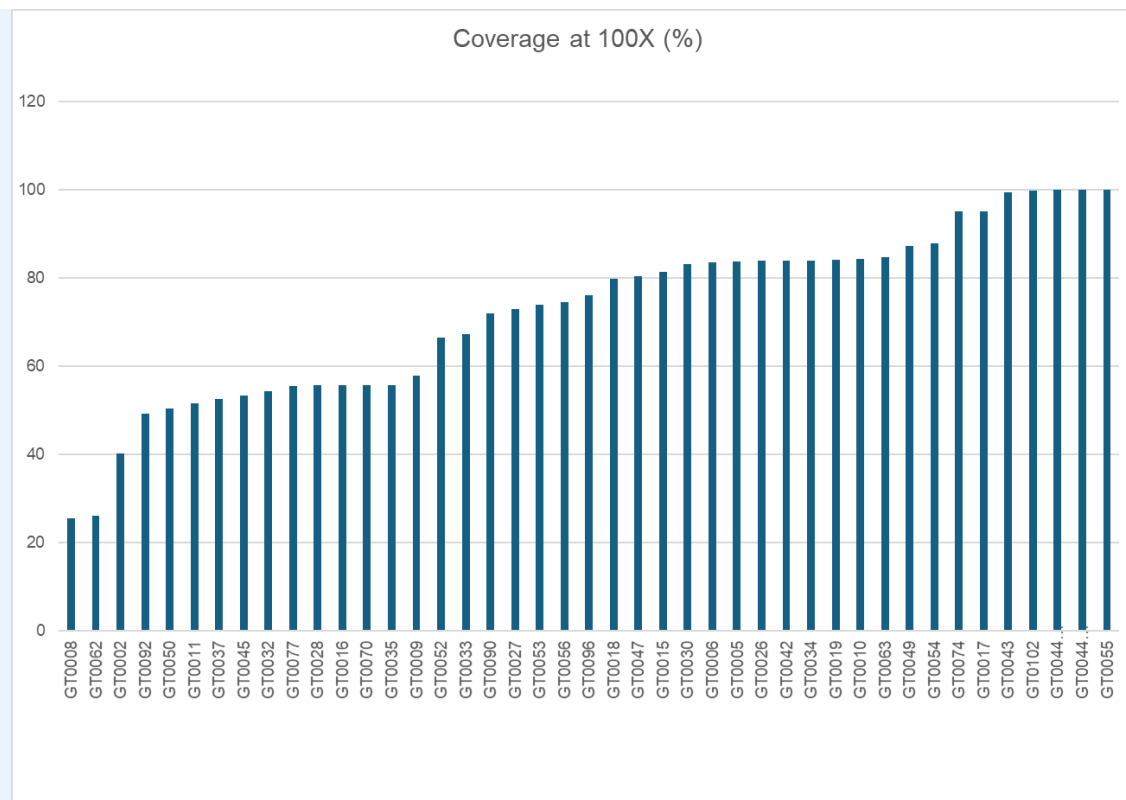
	Moyenne de Read depth (average)
iSEQ 100	5 173
MiniSeq	3 966
NextSeq 500/550	3 283
Genexus	2 783
S5 Ion	1 964
MiSeq	1 531
NextSeq 1000/2000	1 489
Aviti Element bio	1 239

Multiparamétrique – Read depth, Bed à 6 gènes

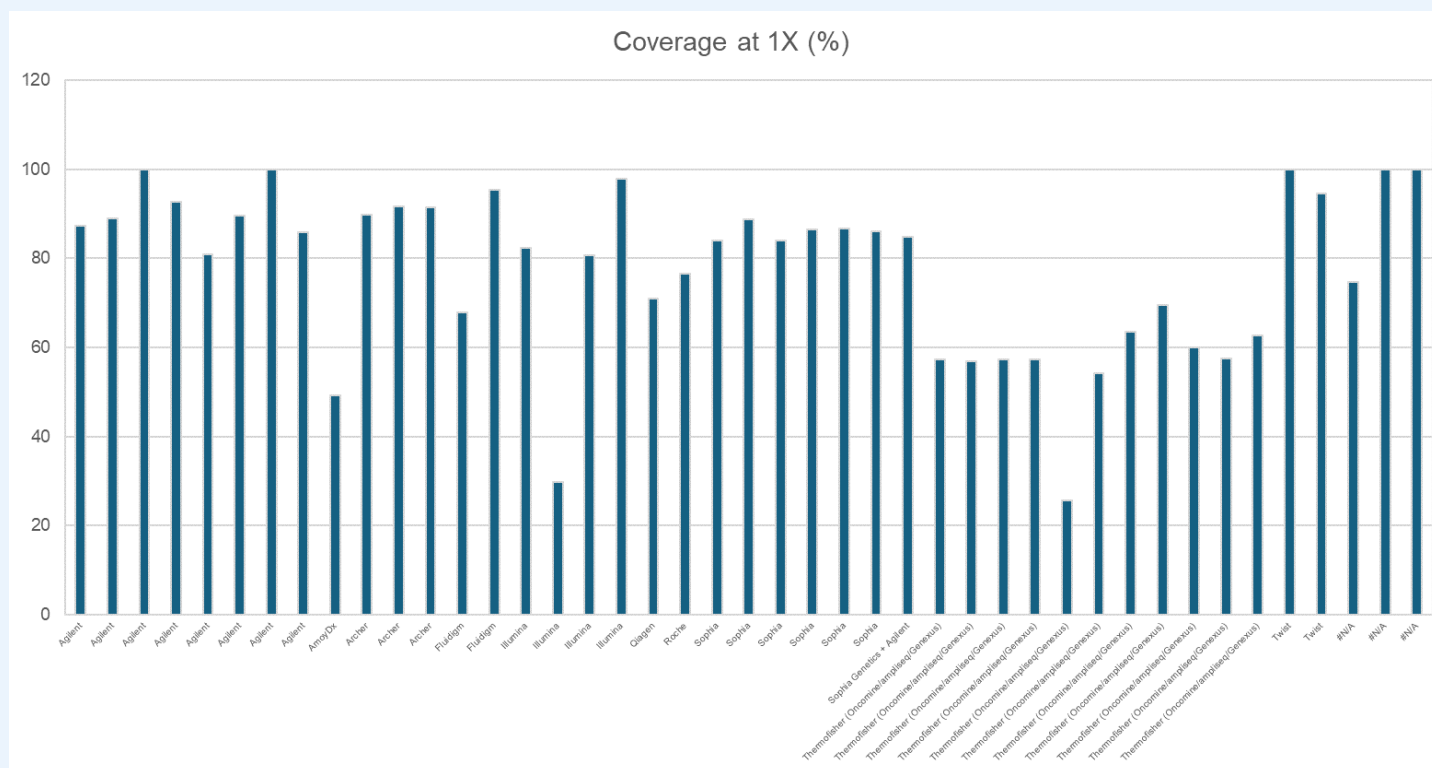


	Moyenne de Error rate on target
iSEQ 100	0,00290
NextSeq 1000/2000	0,00338
MiSeq	0,00463
MiniSeq	0,00522
S5 Ion	0,00614
Aviti Element bio	0,00770
Genexus	0,00835
NextSeq 500/550	0,01085

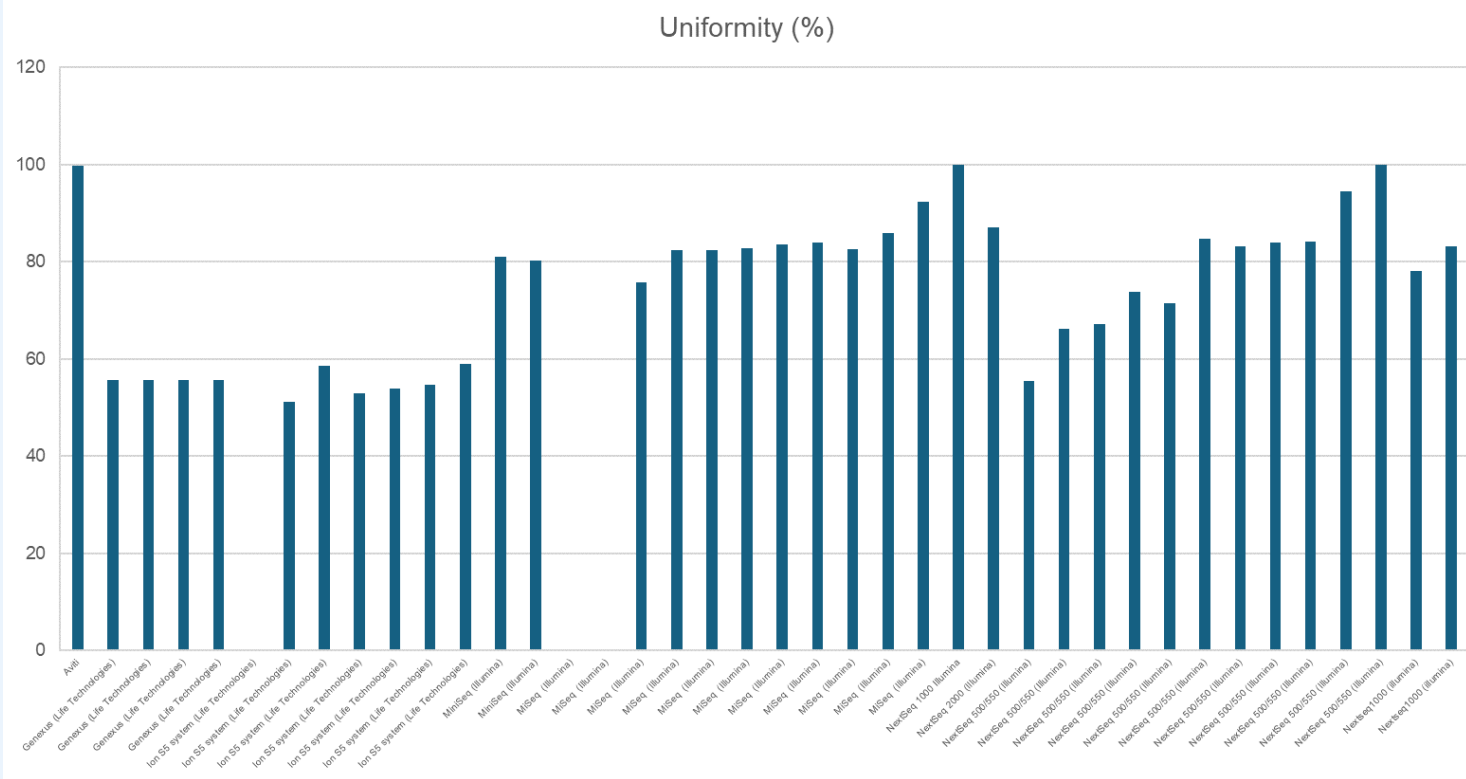
Multiparamétrique – Coverage 100X



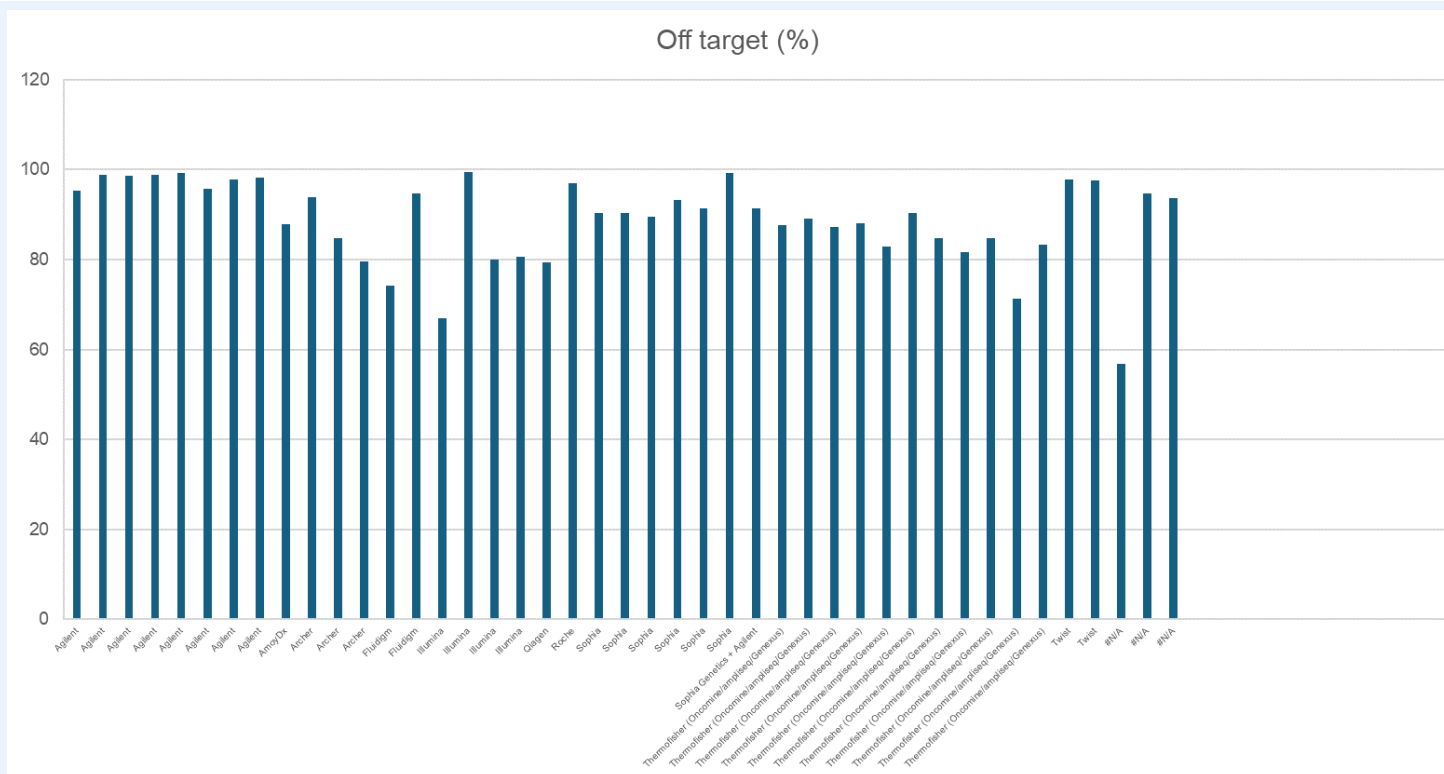
Multiparamétrique – Coverage 1X



Multiparamétrique – Uniformity



Multiparamétrique – Off target



Ovaire – Critères de qualité

	Moyenne Read total
Genexus	1 097 862
NextSeq 2000	1 969 972
Ion S5	2 474 036
Aviti	3 809 342
MiSeq	4 175 979
MiniSeq	6 321 907
NextSeq 1000	11 988 788
NovaSeq	13 896 045
NextSeq 500	18 562 110

	Moyenne de Uniformity (%)
AVITI	100
Genexus	84
Ion S5	84
MiniSeq	89
MiSeq	85
NextSeq 1000	95
NextSeq 2000	91
NextSeq 500	92
NovaSeq	82

	Moyenne de Base quality (average)
Aviti	81
MiniSeq	39
MiSeq	38
NovaSeq	37
NextSeq 500	35
NextSeq 1000	34
NextSeq 2000	33
Ion S5	30
Genexus	24

	Moyenne de Off target (%)
NovaSeq	99
NextSeq 2000	96
NextSeq 500	91
MiniSeq	91
NextSeq 1000	70
MiSeq	68
Ion S5	33
AVITI	16
Genexus	7

	Moyenne de Read depth (average)
Genexus	3 204
Ion S5	3 106
MiniSeq	2 638
NextSeq 500	2 216
AVITI	1 561
MiSeq	1 453
NextSeq 1000	1 322
NextSeq 2000	380
NovaSeq	342

	Moyenne de Reads mapped (%)
NextSeq 2000	100
NovaSeq	100
MiniSeq	100
NextSeq 1000	100
NextSeq 500	100
MiSeq	99
AVITI	99
Ion S5	98
Genexus	93

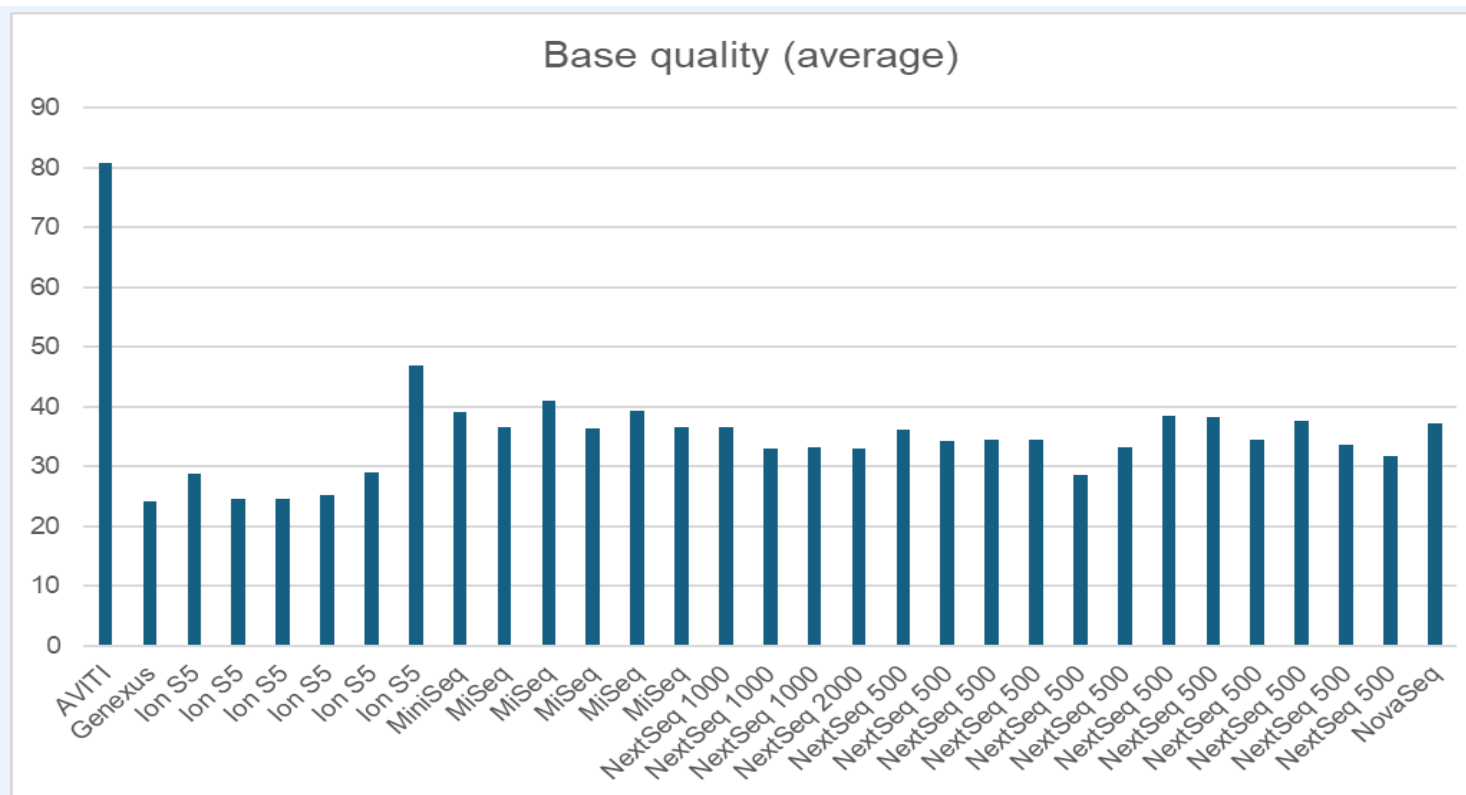
Ovaire – Bilan des critères de qualité

	Moyenne de Coverage at 1X (%)
AVITI	100
NextSeq 1000	100
NextSeq 2000	95
NextSeq 500	94
MiniSeq	92
MiSeq	88
Ion S5	87
Genexus	86
NovaSeq	85

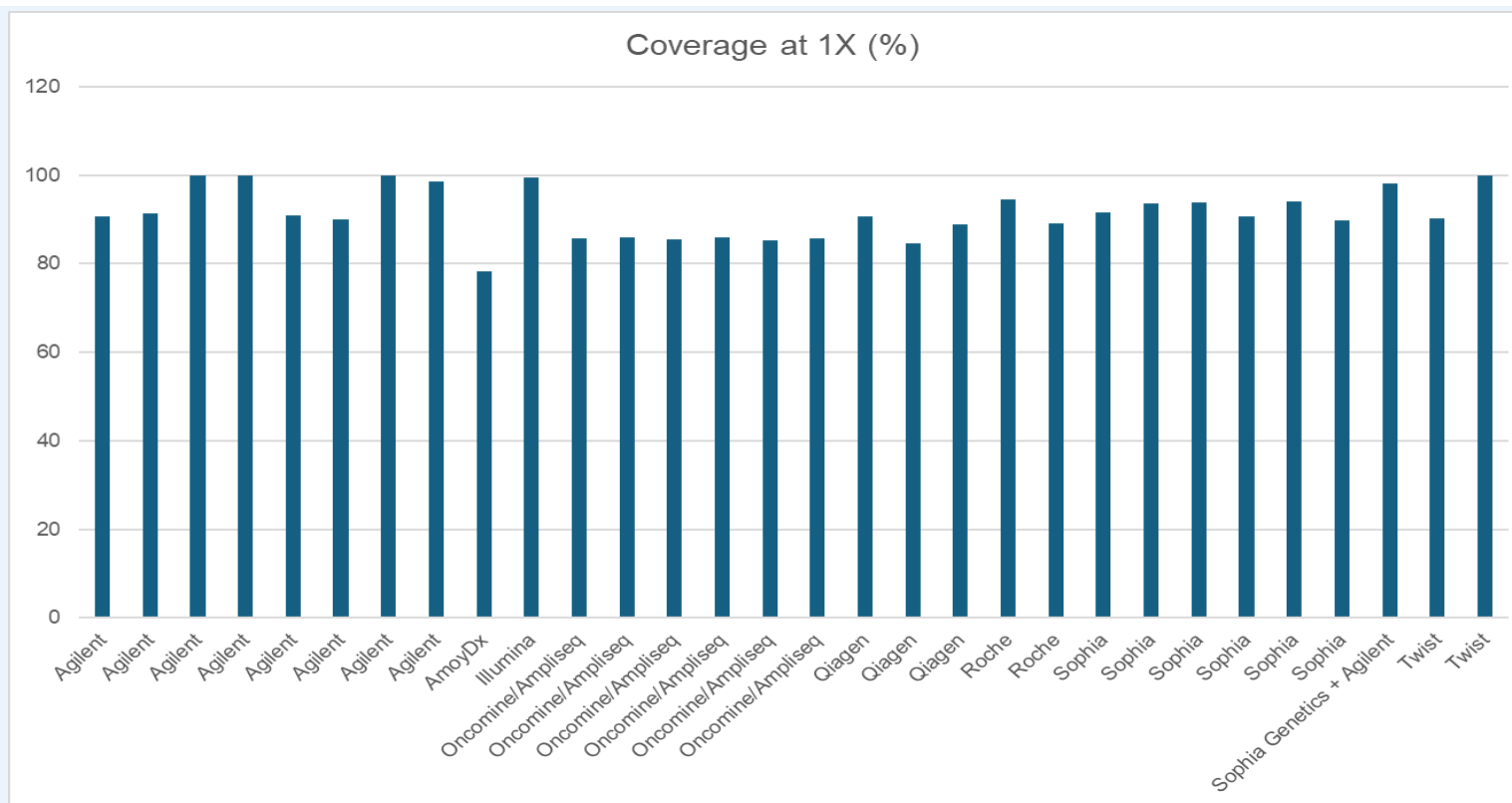
	Moyenne de Coverage at 100X (%)
AVITI	100
NextSeq 1000	93
NextSeq 500	92
MiniSeq	89
NextSeq 2000	86
MiSeq	86
Ion S5	85
Genexus	84
NovaSeq	76

	Moyenne de Ti/Tv
AVITI	14
NextSeq 2000	6
Genexus	6
MiniSeq	5
MiSeq	3
NovaSeq	3
Ion S5	3
NextSeq 500	2
NextSeq 1000	2

Ovaire – Base quality

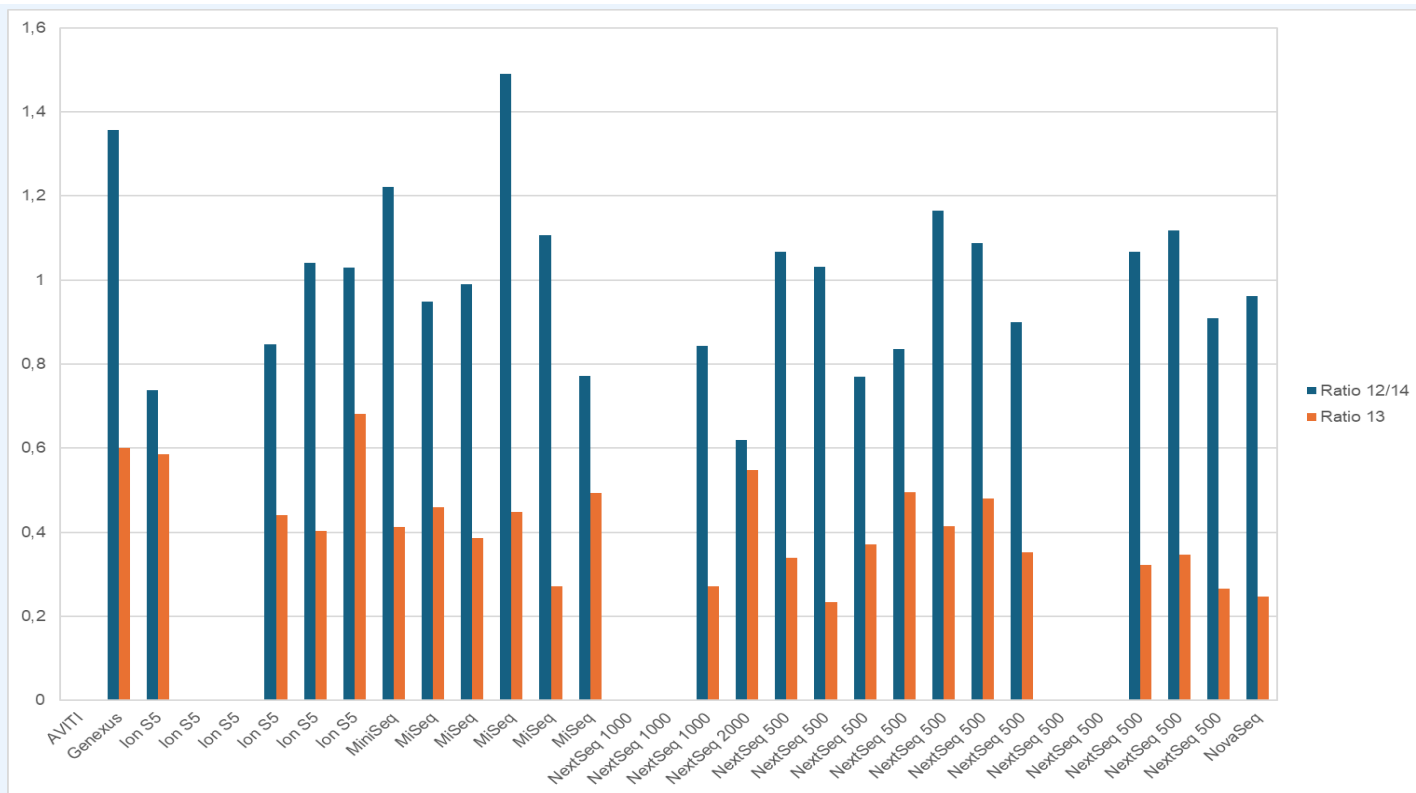


Ovaire – Coverage 1X



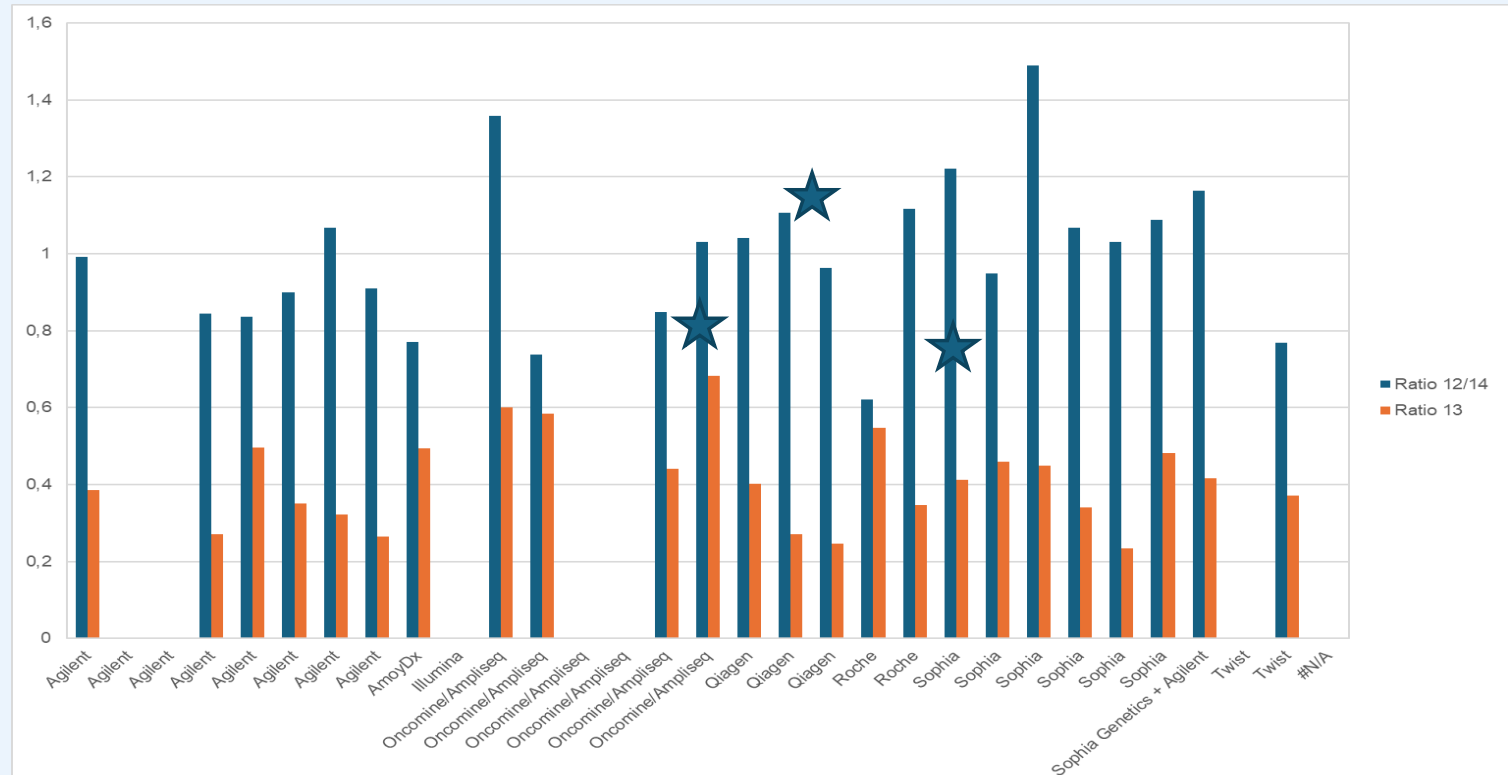
Ovaire – Impact de la technique sur le ratio exon 12-13-14

Par séquenceur



Impact de la technique sur le ratio exon 12-13-14

Par librairie



Conclusion

- Données encore en cours de consolidation
- Analyse des variants en cours
- Bilan en fonction des paires
- Importance de l'identification des techniques
- Approche sur un bed unique: limites et apports
- Critères discriminants à mieux déterminer



Contacts

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