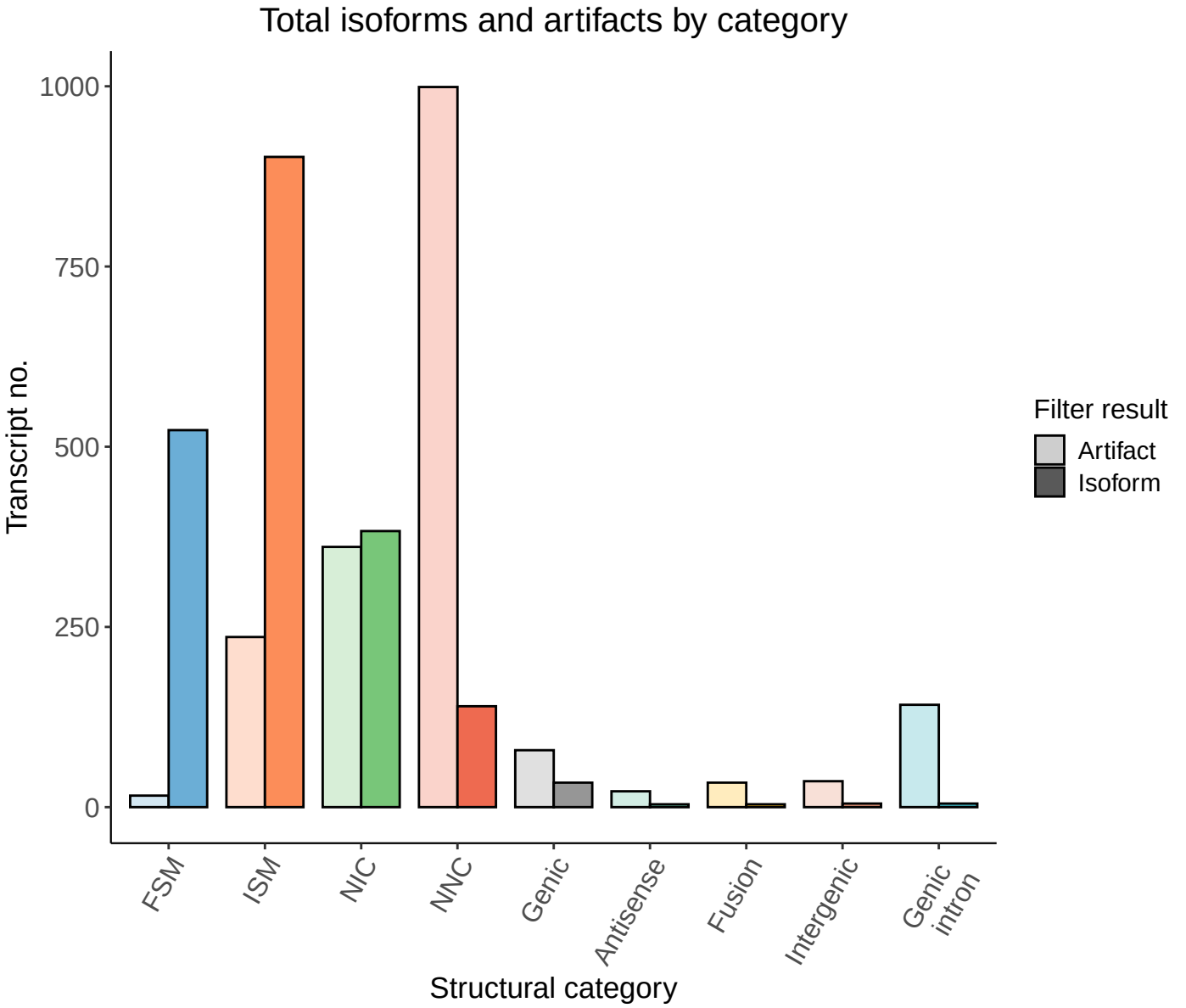
# *SQANTI3 filter report*

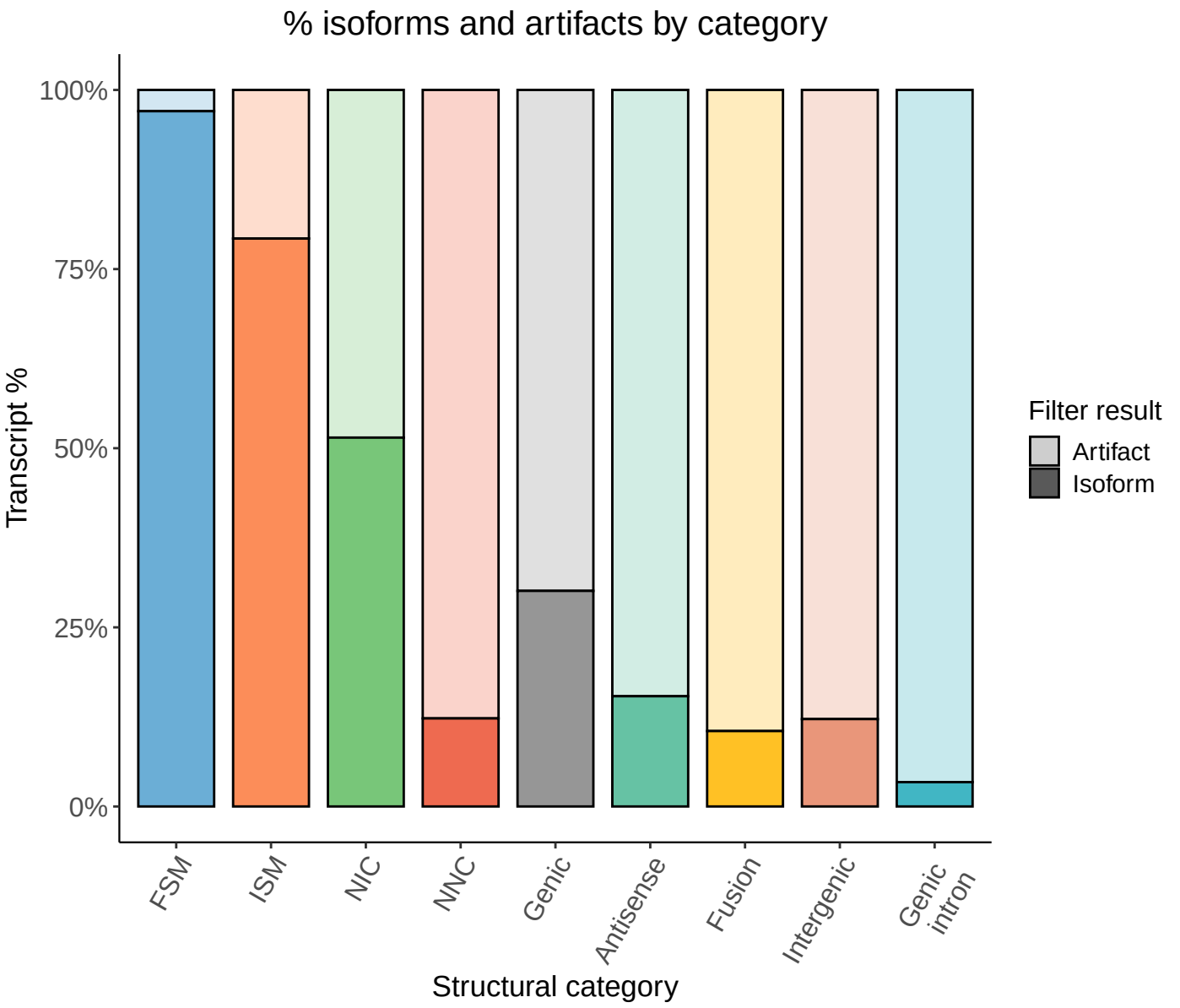
*Total Genes: 656*

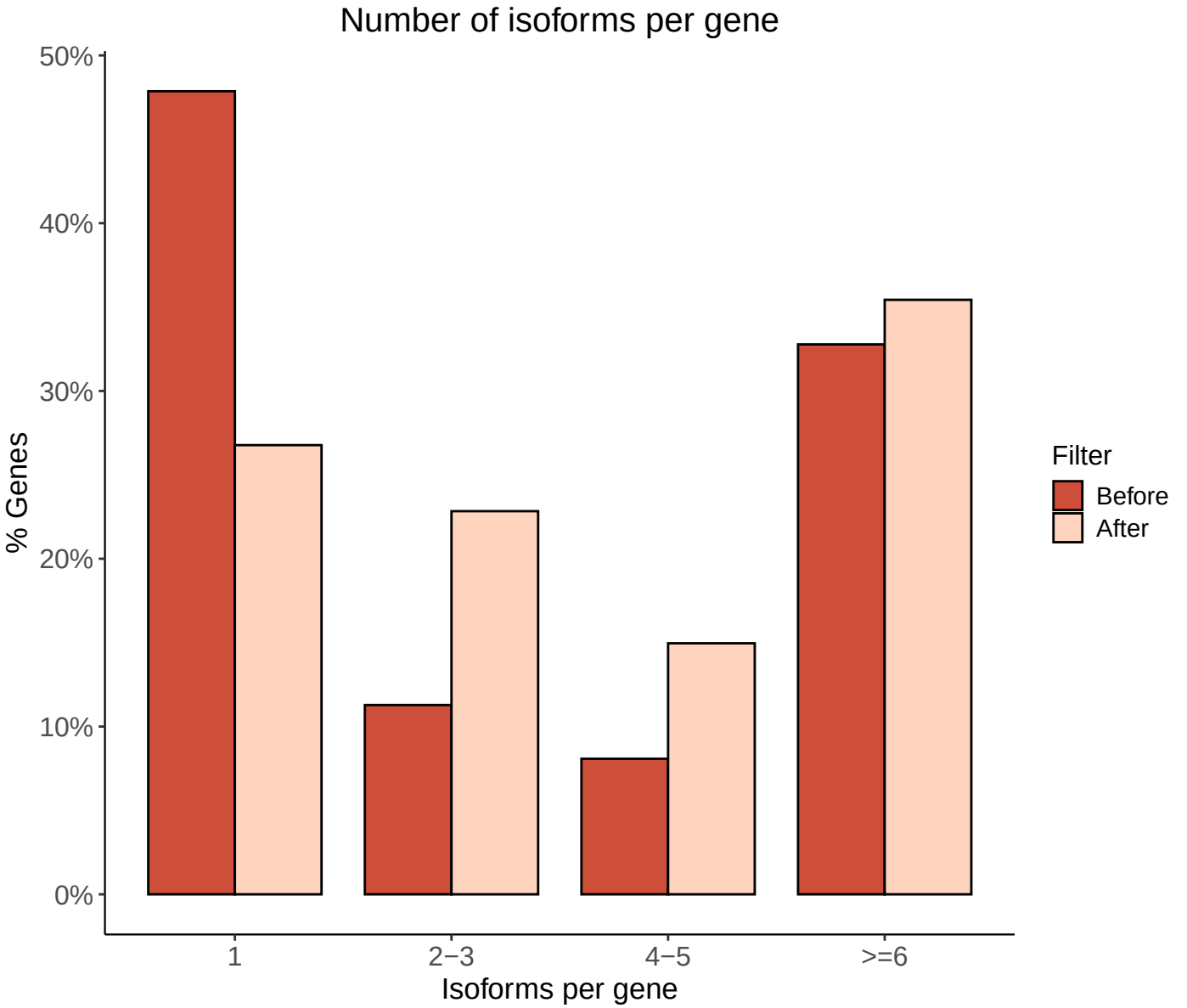
*Total Transcripts: 3925*
*− Isoforms: 2000 (51%)*
*− Artifacts: 1925 (49%)*

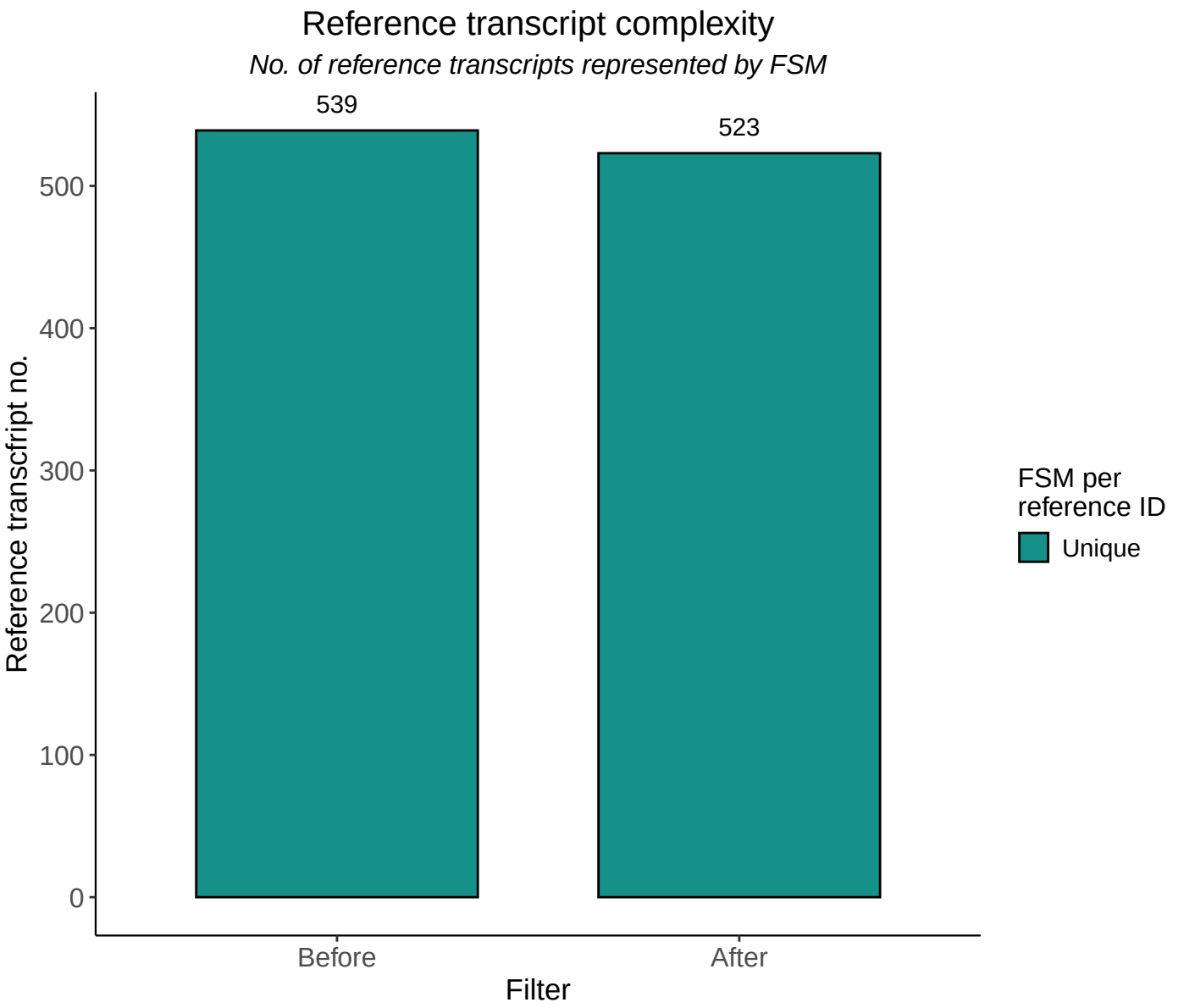
| Gene category | Gene no. | No. of genes with artifacts only |
|---|---|---|
| Annotated | 443 | 76 |
| Novel | 213 | 199 |

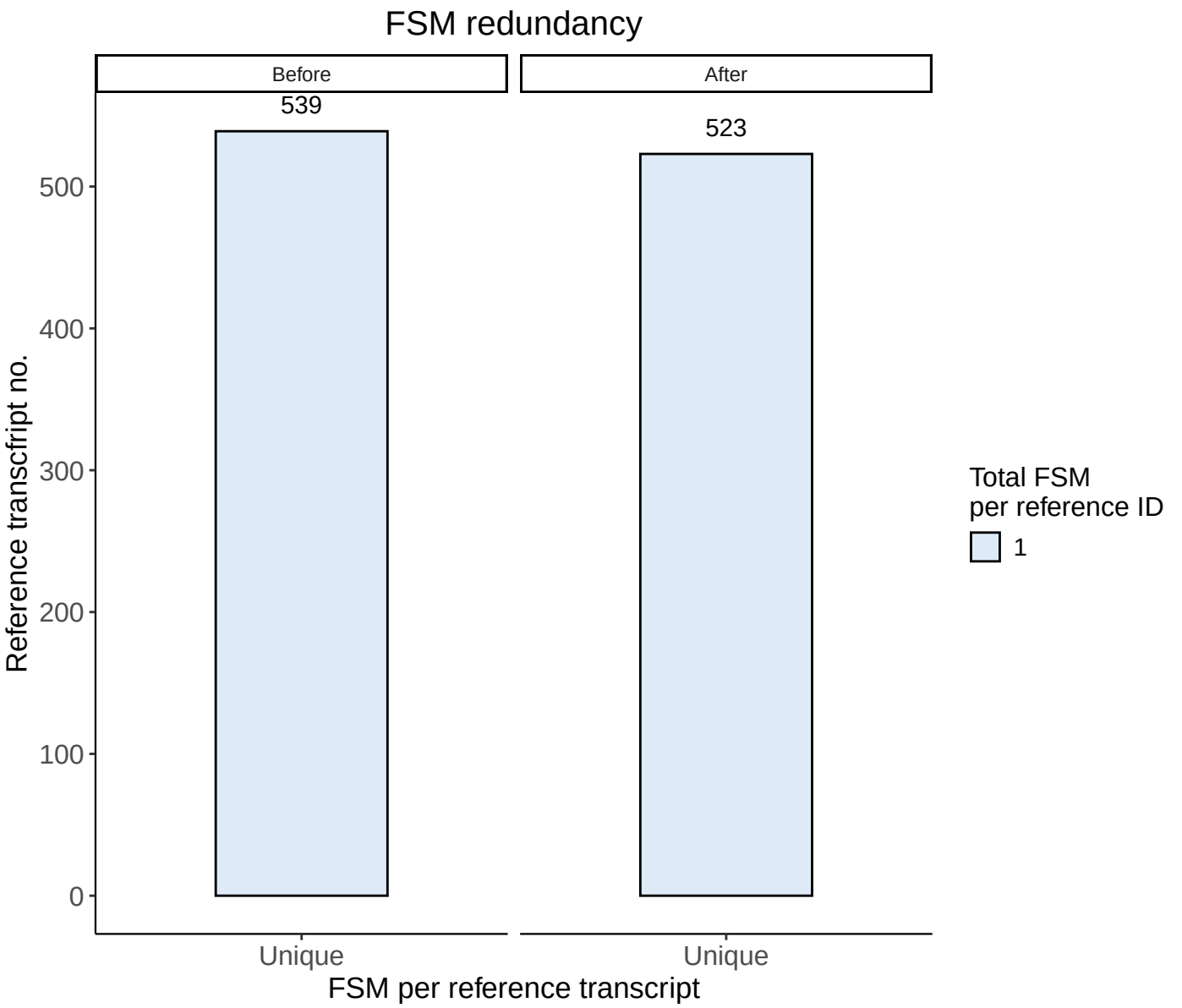
| Structural category | Artifact no. | Isoform no. |
|---|---|---|
| FSM | 16 | 523 |
| ISM | 236 | 902 |
| NIC | 361 | 383 |
| NNC | 999 | 140 |
| Genic | 79 | 34 |
| Antisense | 22 | 4 |
| Fusion | 34 | 4 |
| Intergenic | 36 | 5 |
| Genic intron | 142 | 5 |

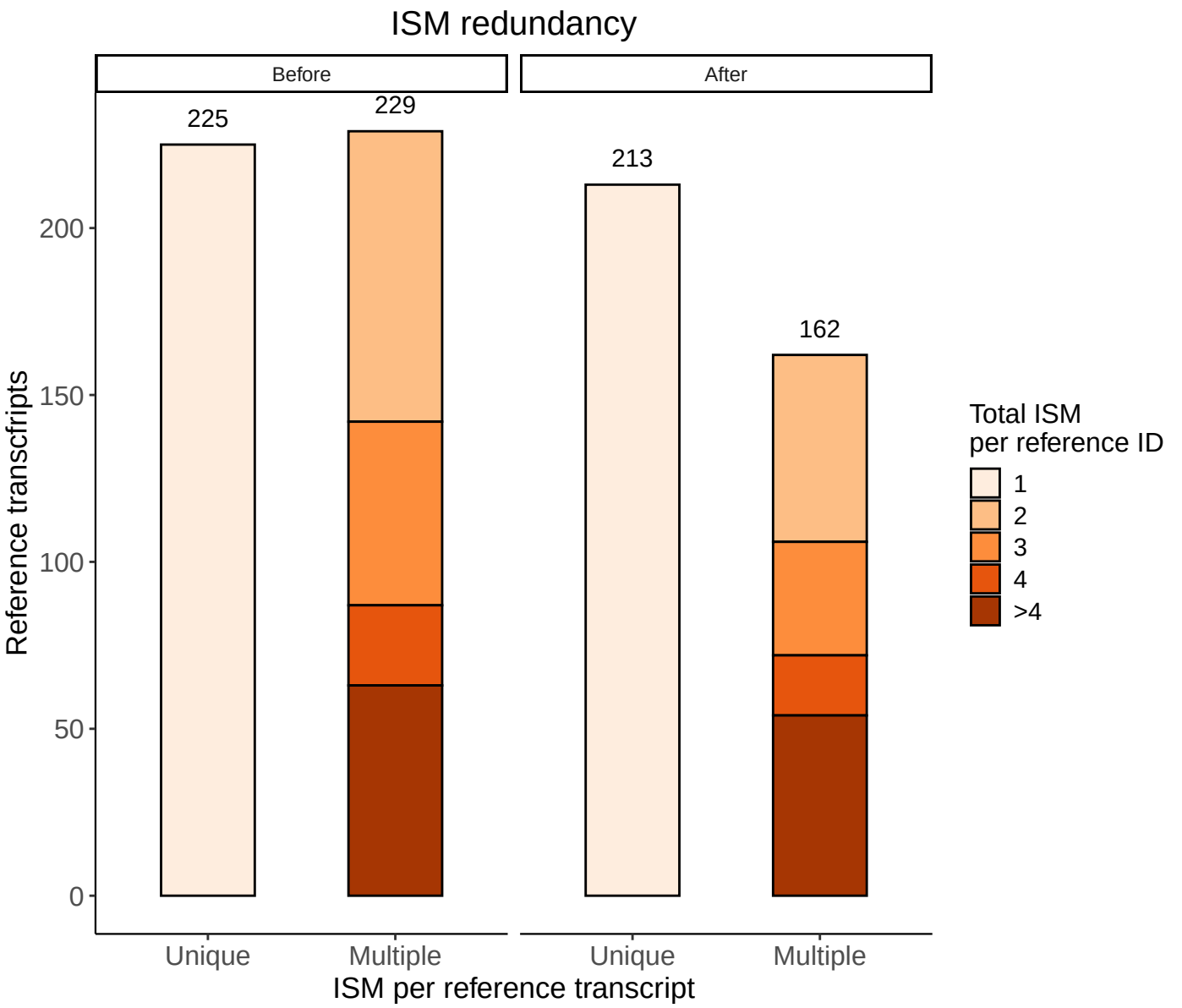

Total isoforms and artifacts by category
Transcript no.
1000
750
500
250
0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

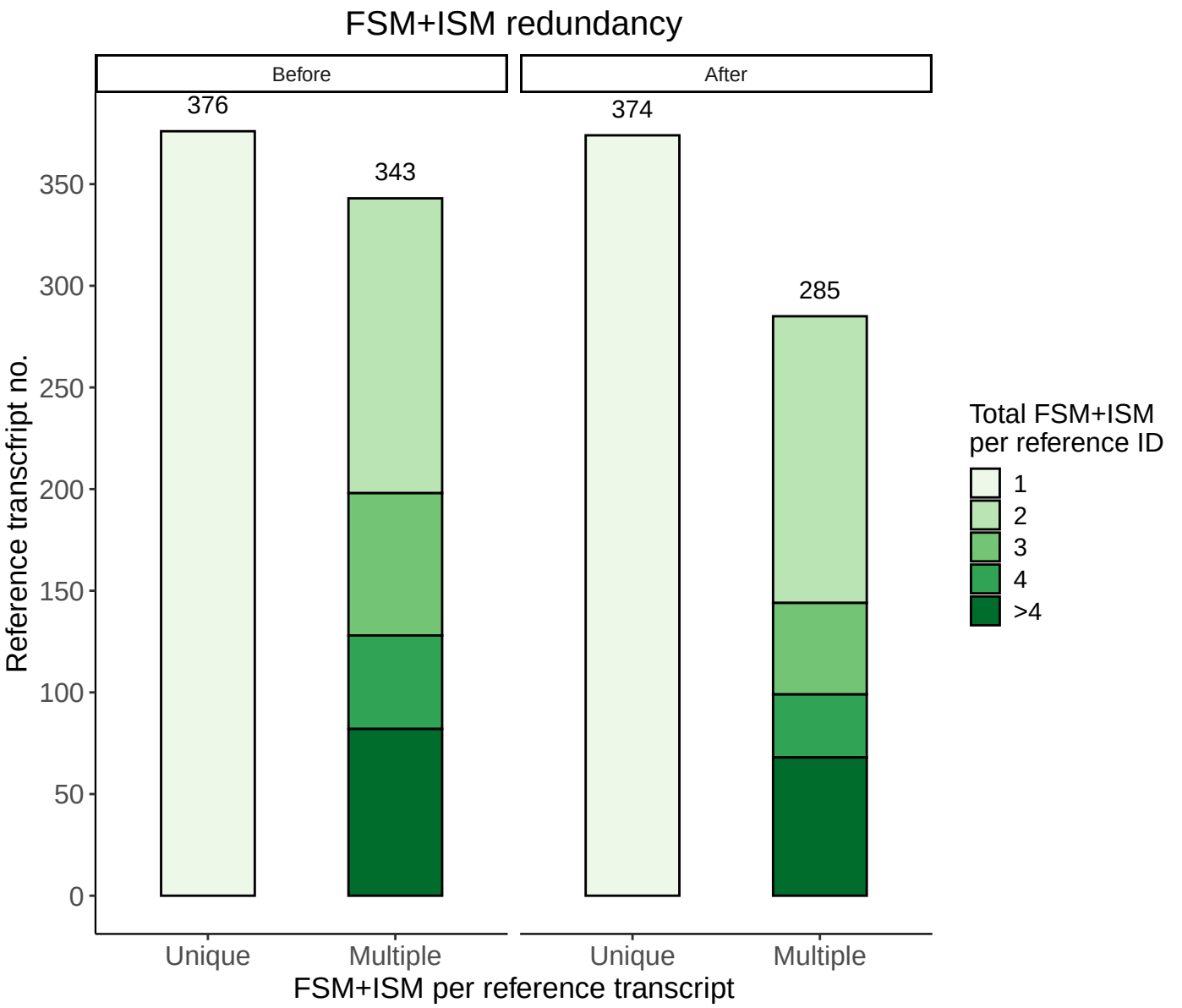

% isoforms and artifacts by category
100%
75%
50%
25%
0%
Transcript %
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform


Number of isoforms per gene
50%
40%
30%
20%
10%
0%
% Genes
1
2−3
4−5
>=6
Isoforms per gene
Filter
Before
After


Reference transcript complexity
No. of reference transcripts represented by FSM
539
523
500
400
300
200
100
0
Reference transcfript no.
Before
After
Filter
FSM per reference ID
Unique


FSM redundancy
Before
After
539
523
500
400
300
200
100
0
Reference transcfript no.
Unique
Unique
FSM per reference transcript
Total FSM
per reference ID
1


ISM redundancy
Before
After
225
229
213
162
200
150
100
50
0
Reference transcfripts
Unique
Multiple
Unique
Multiple
ISM per reference transcript
Total ISM
per reference ID
1
2
3
4
>4


FSM+ISM redundancy
Before
After
376
343
374
285
350
300
250
200
150
100
50
0
Reference transcfript no.
Unique
Multiple
Unique
Multiple
FSM+ISM per reference transcript
Total FSM+ISM per reference ID
1
2
3
4
>4

# *ML classifier performance report*

# Classification model performance on test set

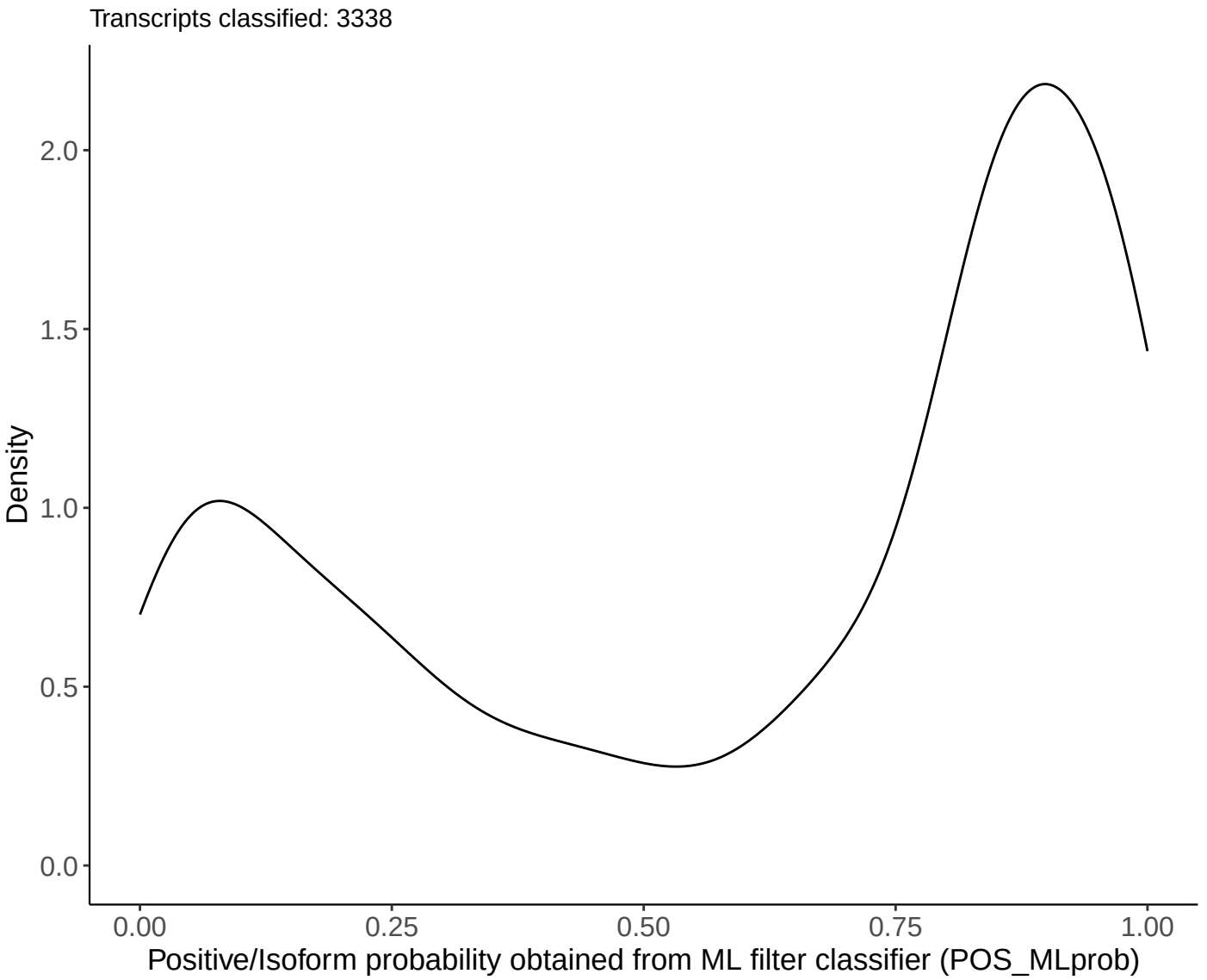
## Performance metrics

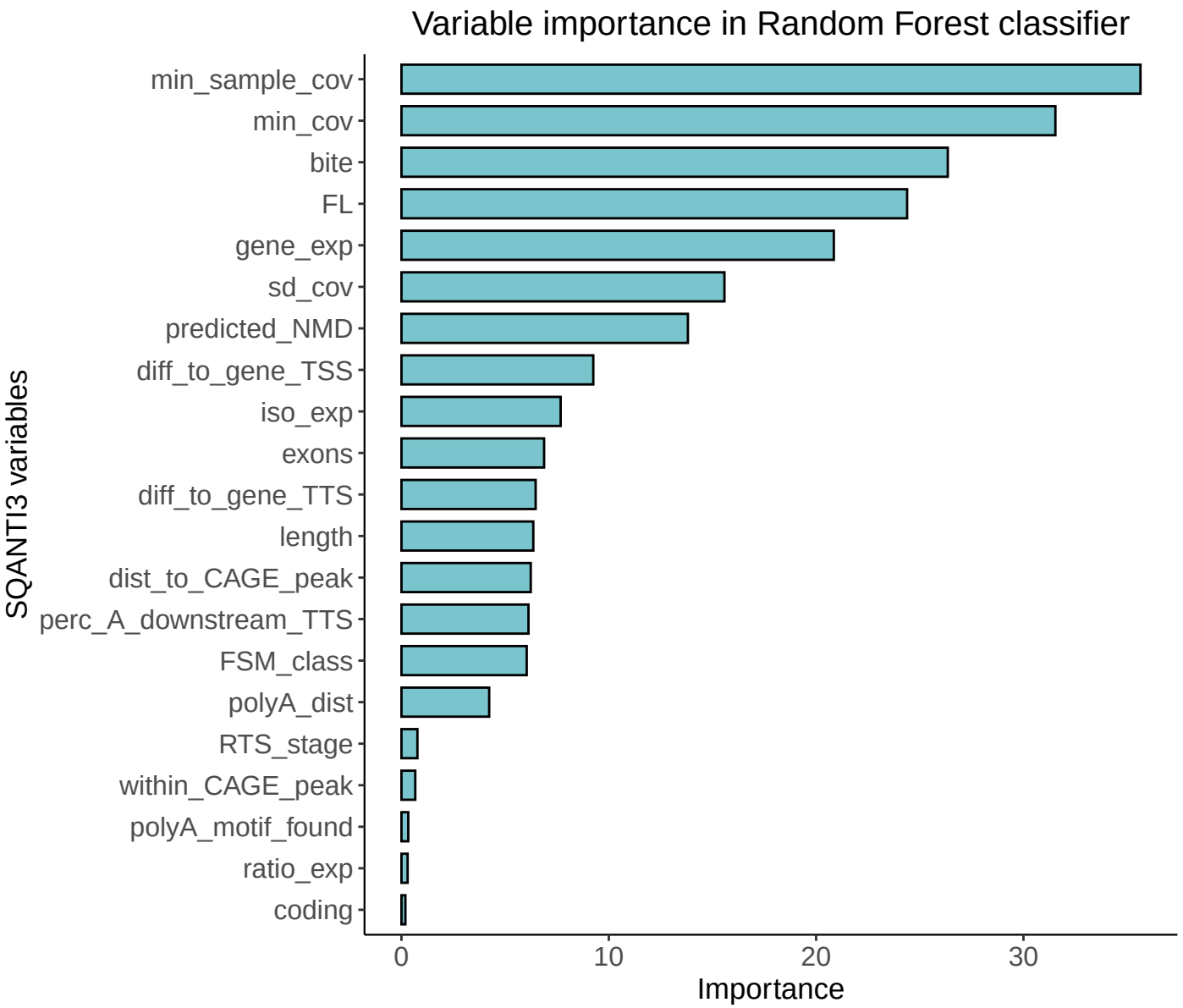
| Metric | Value |
|---|---|
| Accuracy | 0.904 |
| Kappa | 0.807 |
| AccuracyLower | 0.834 |
| AccuracyUpper | 0.951 |
| AccuracyNull | 0.5 |
| McnemarPValue | 0.0159 |
| Sensitivity | 0.825 |
| Specificity | 0.982 |
| Pos Pred Value | 0.979 |
| Neg Pred Value | 0.848 |
| Precision | 0.979 |
| Recall | 0.825 |
| F1 | 0.895 |
| Prevalence | 0.5 |
| Detection Rate | 0.412 |
| Detection Prevalence | 0.421 |
| Balanced Accuracy | 0.904 |

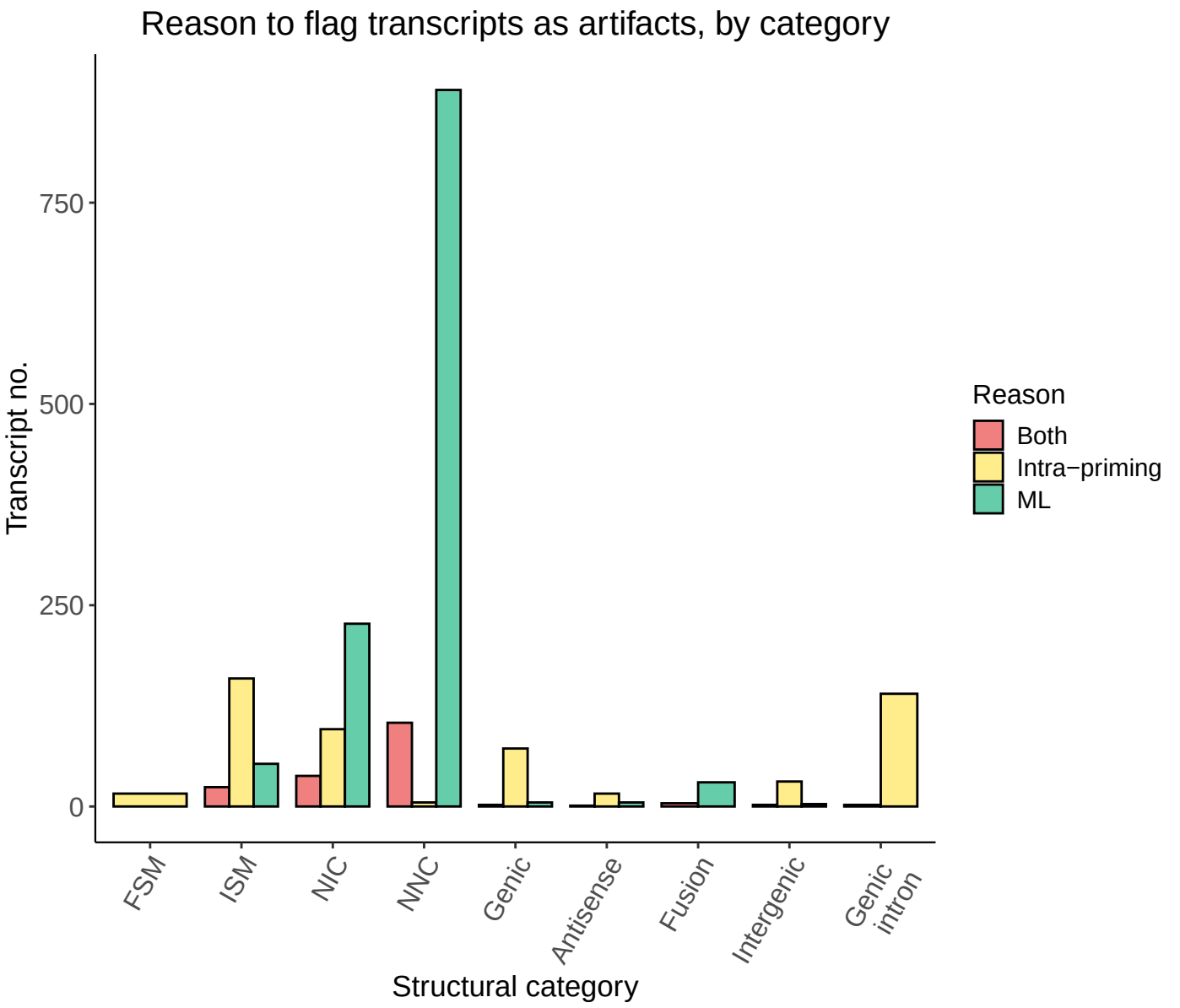
| | |
|---|---|
| AccuracyPValue | 3.459924e−20 |

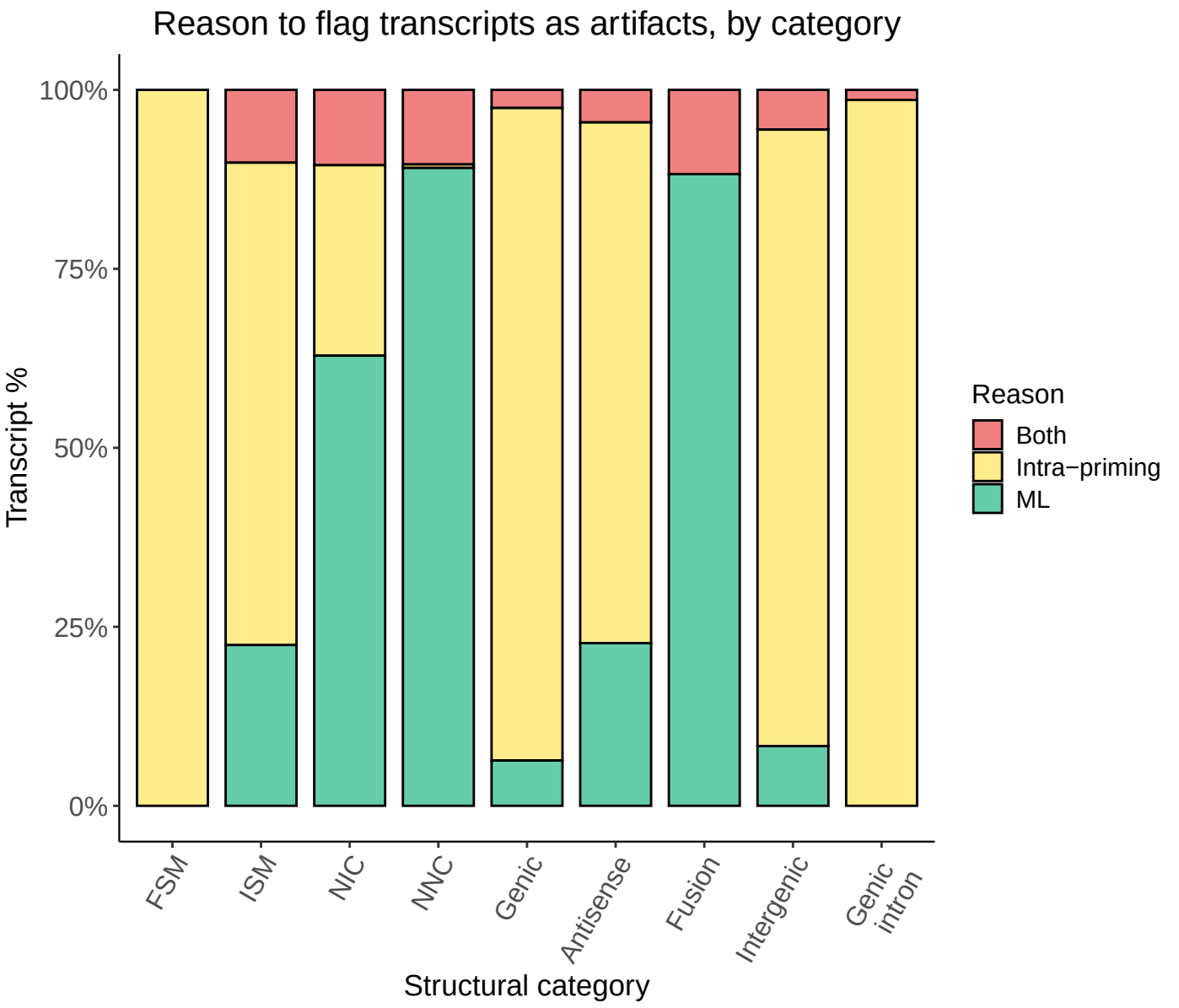
## Confusion matrix

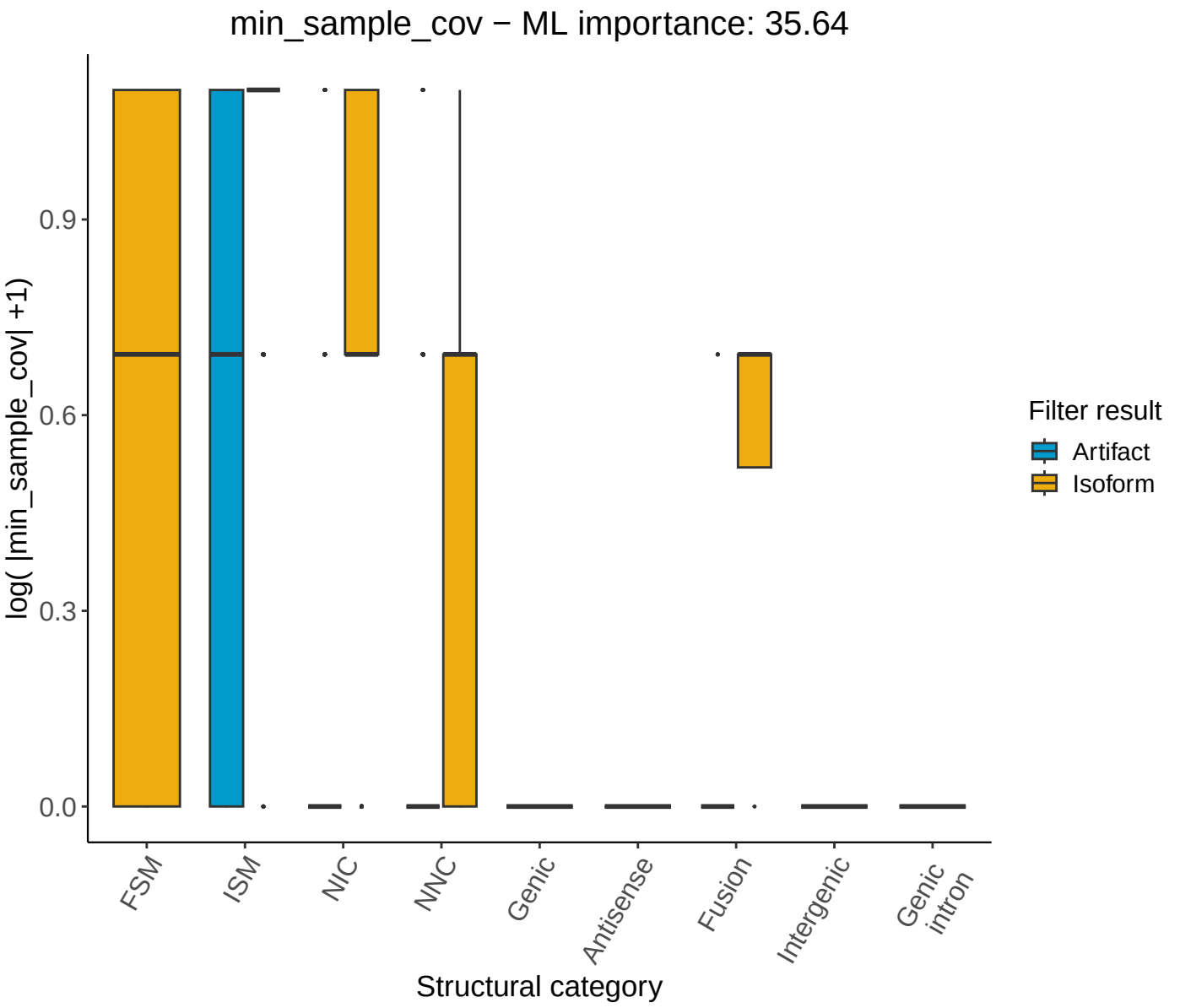
| Prediction | Reference | Freq |
|---|---|---|
| POS | POS | 47 |
| NEG | POS | 10 |
| POS | NEG | 1 |
| NEG | NEG | 56 |

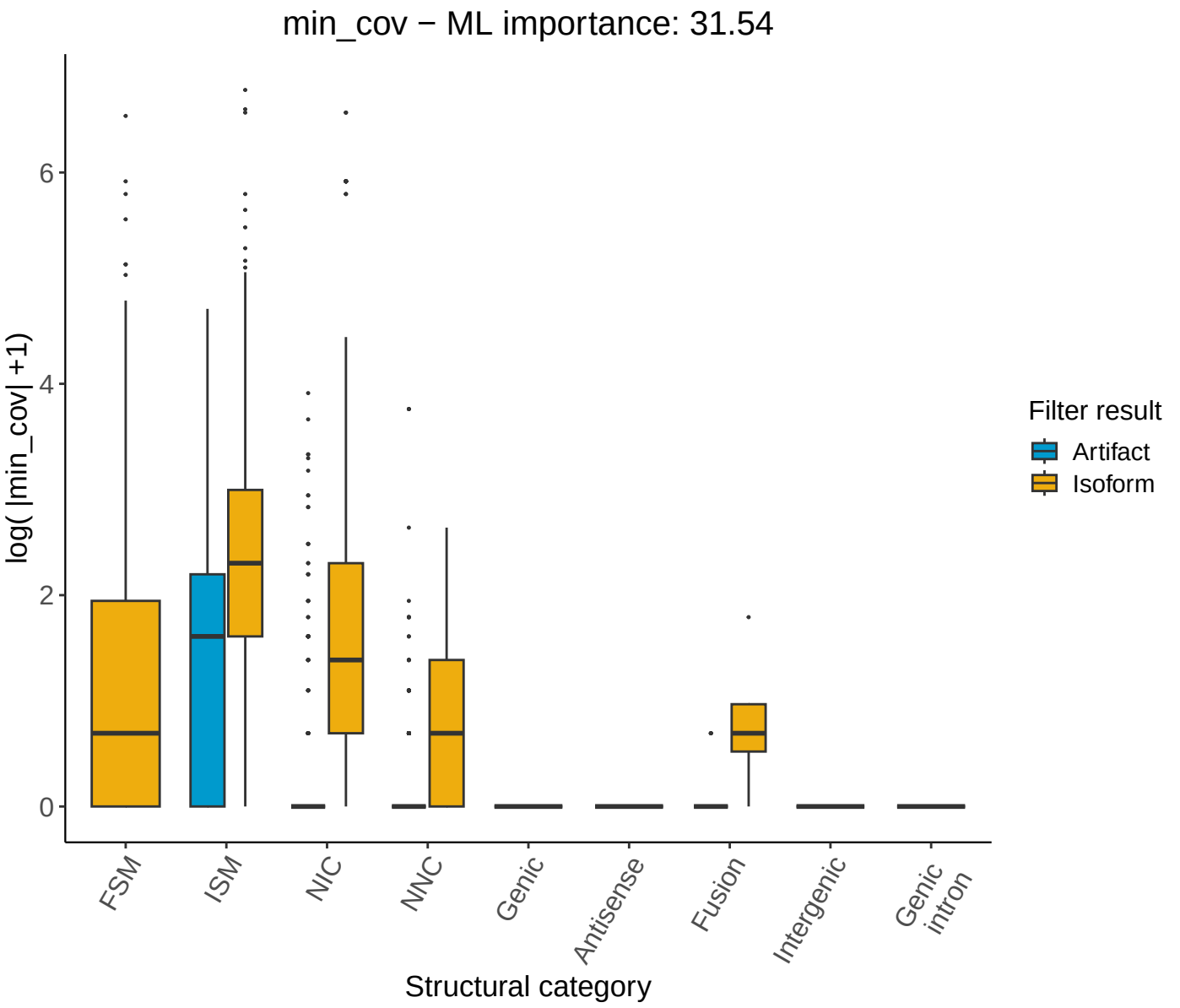

Transcripts classified: 3338
Density
2.0
1.5
1.0
0.5
0.0
0.00
0.25
0.50
0.75
1.00
Positive/Isoform probability obtained from ML filter classifier (POS_MLprob)

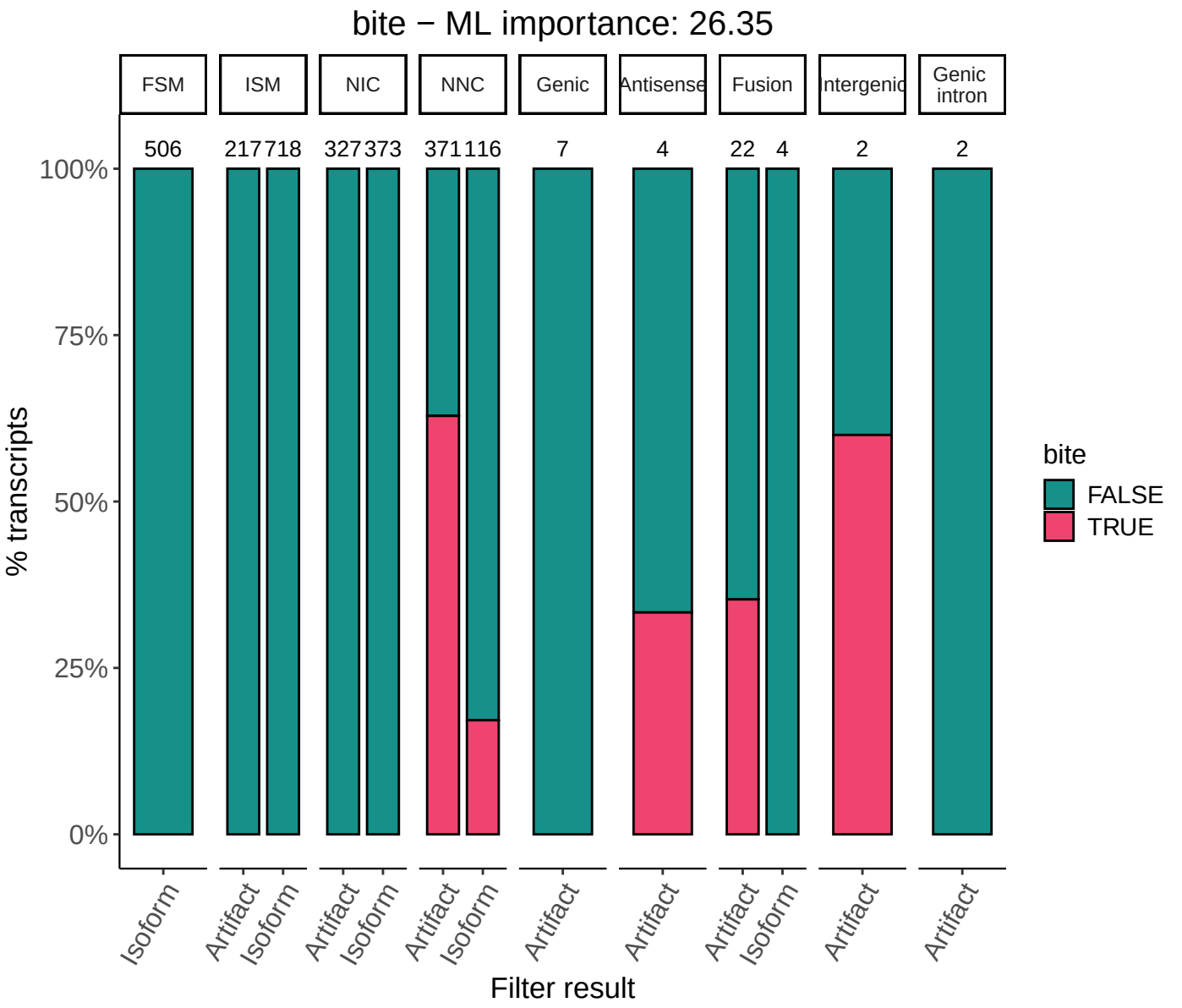

Variable importance in Random Forest classifier
SQANTI3 variables
min_sample_cov
min_cov
bite
FL
gene_exp
sd_cov
predicted_NMD
diff_to_gene_TSS
iso_exp
exons
diff_to_gene_TTS
length
dist_to_CAGE_peak
perc_A_downstream_TTS
FSM_class
polyA_dist
RTS_stage
within_CAGE_peak
polyA_motif_found
ratio_exp
coding
0
10
20
30
Importance

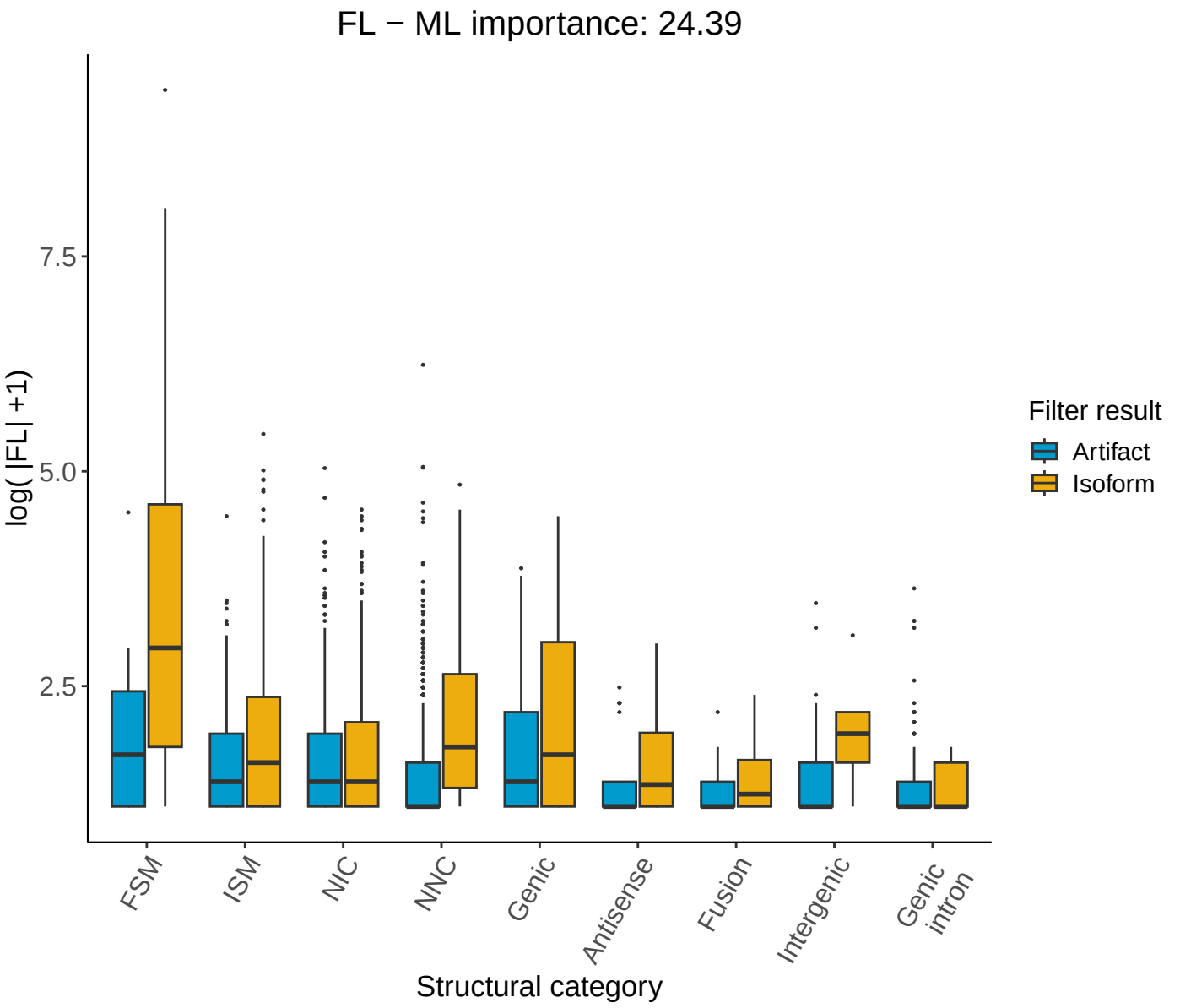

Reason to flag transcripts as artifacts, by category
Transcript no.
750
500
250
0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Reason
Both
Intra−priming
ML

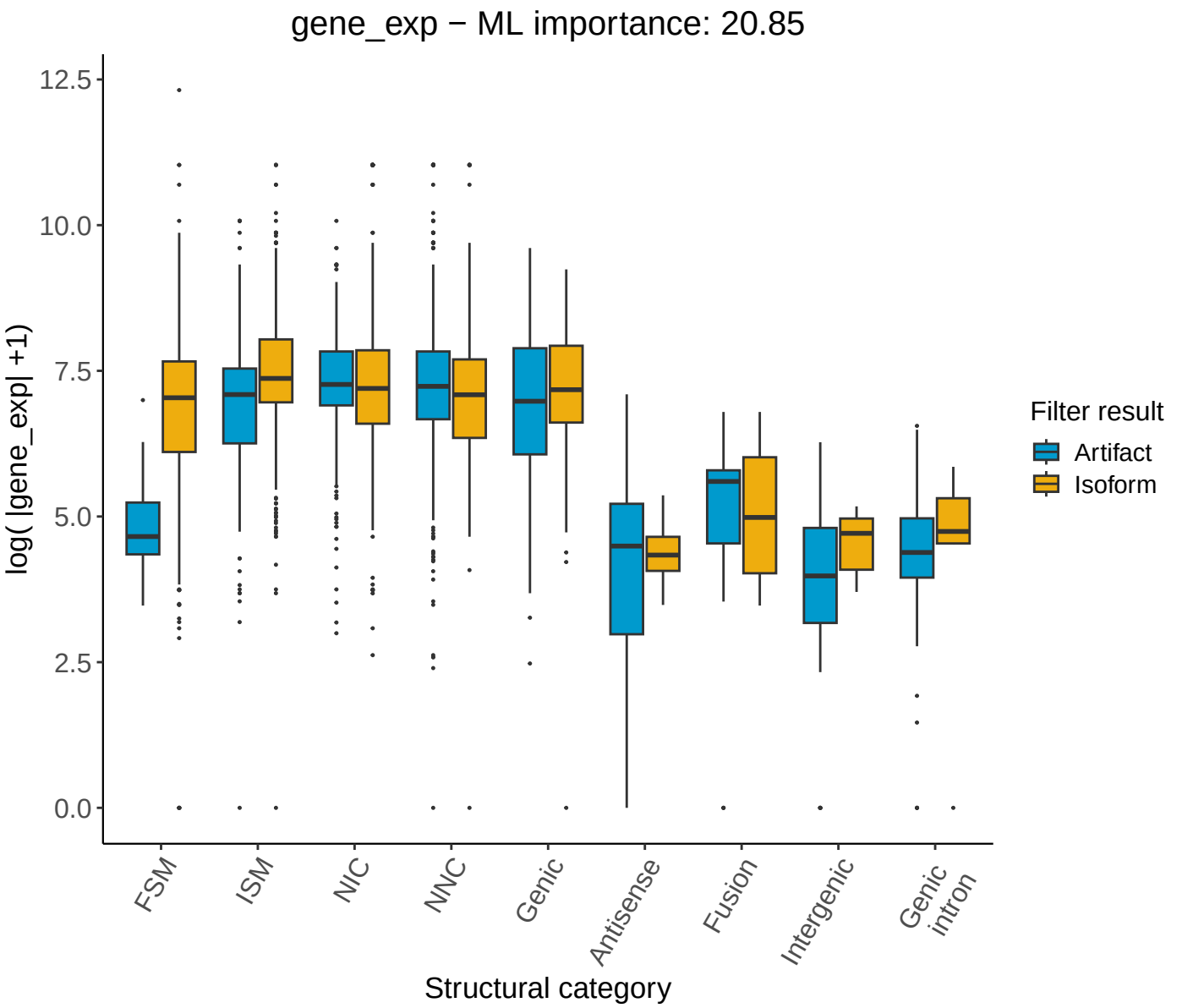

Reason to flag transcripts as artifacts, by category
100%
75%
50%
25%
0%
Transcript %
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Reason
Both
Intra−priming
ML

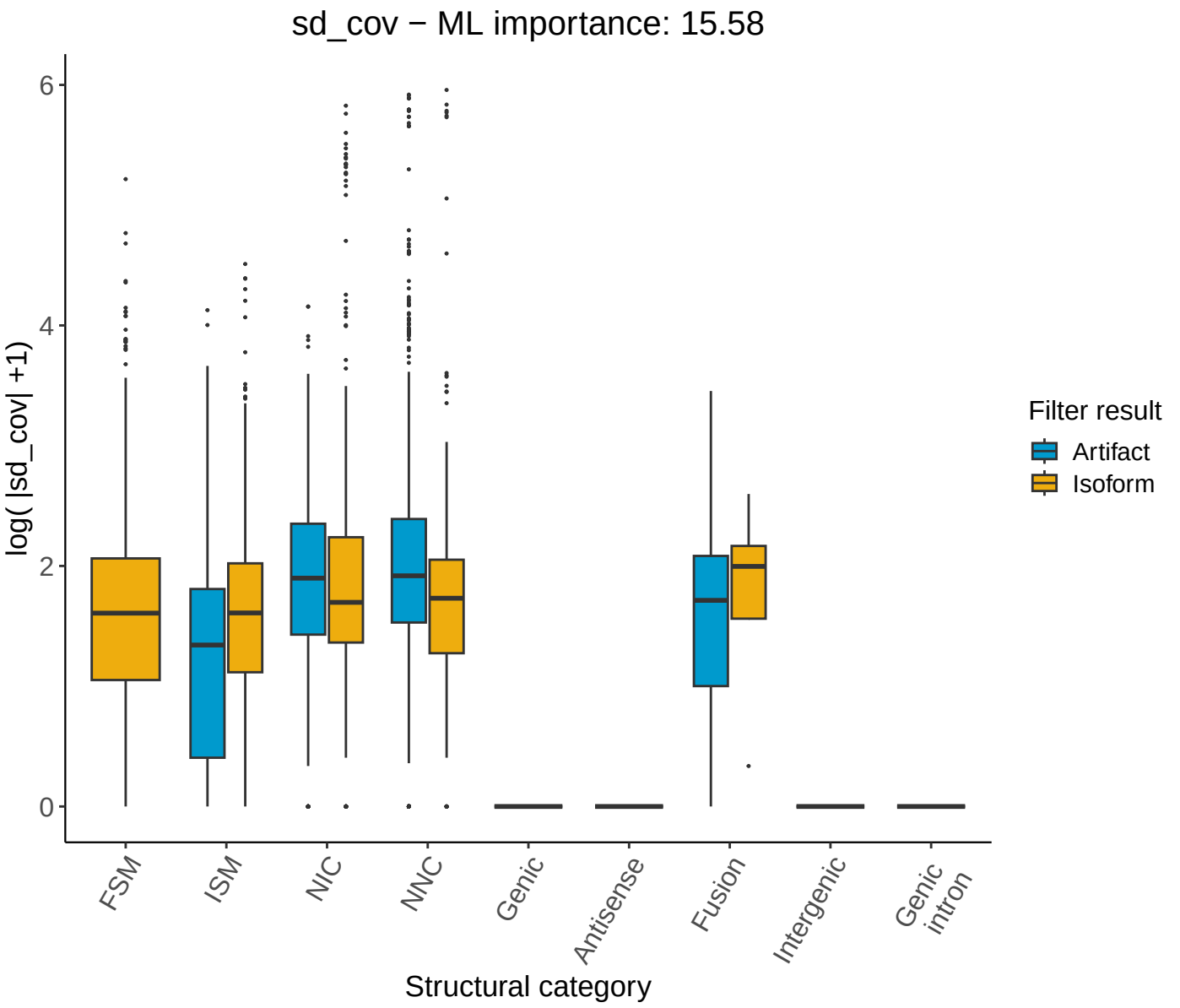

min_sample_cov − ML importance: 35.64
log( |min_sample_cov| +1)
0.0
0.3
0.6
0.9
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

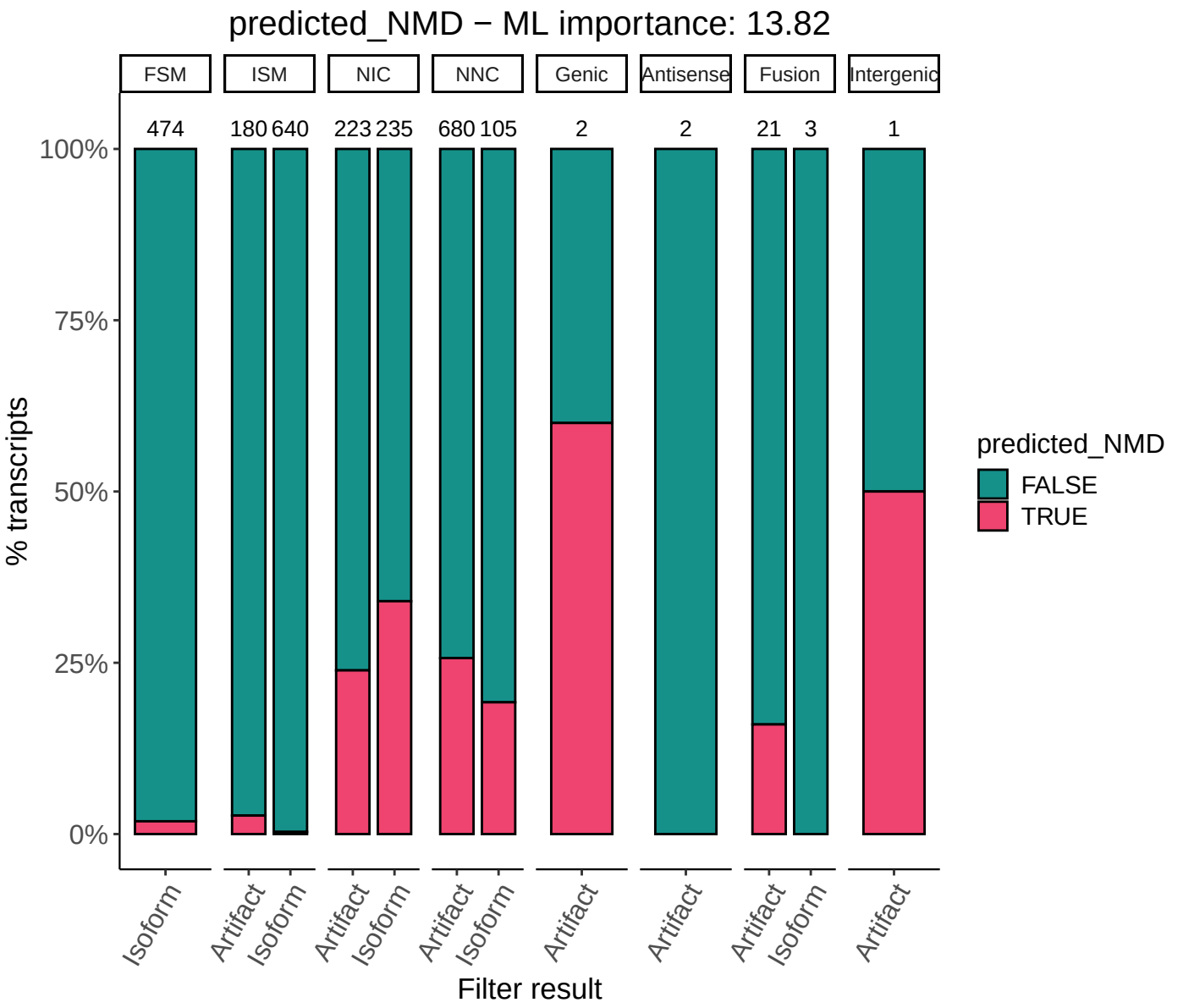

min_cov − ML importance: 31.54
log( |min_cov| +1)
6
4
2
0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

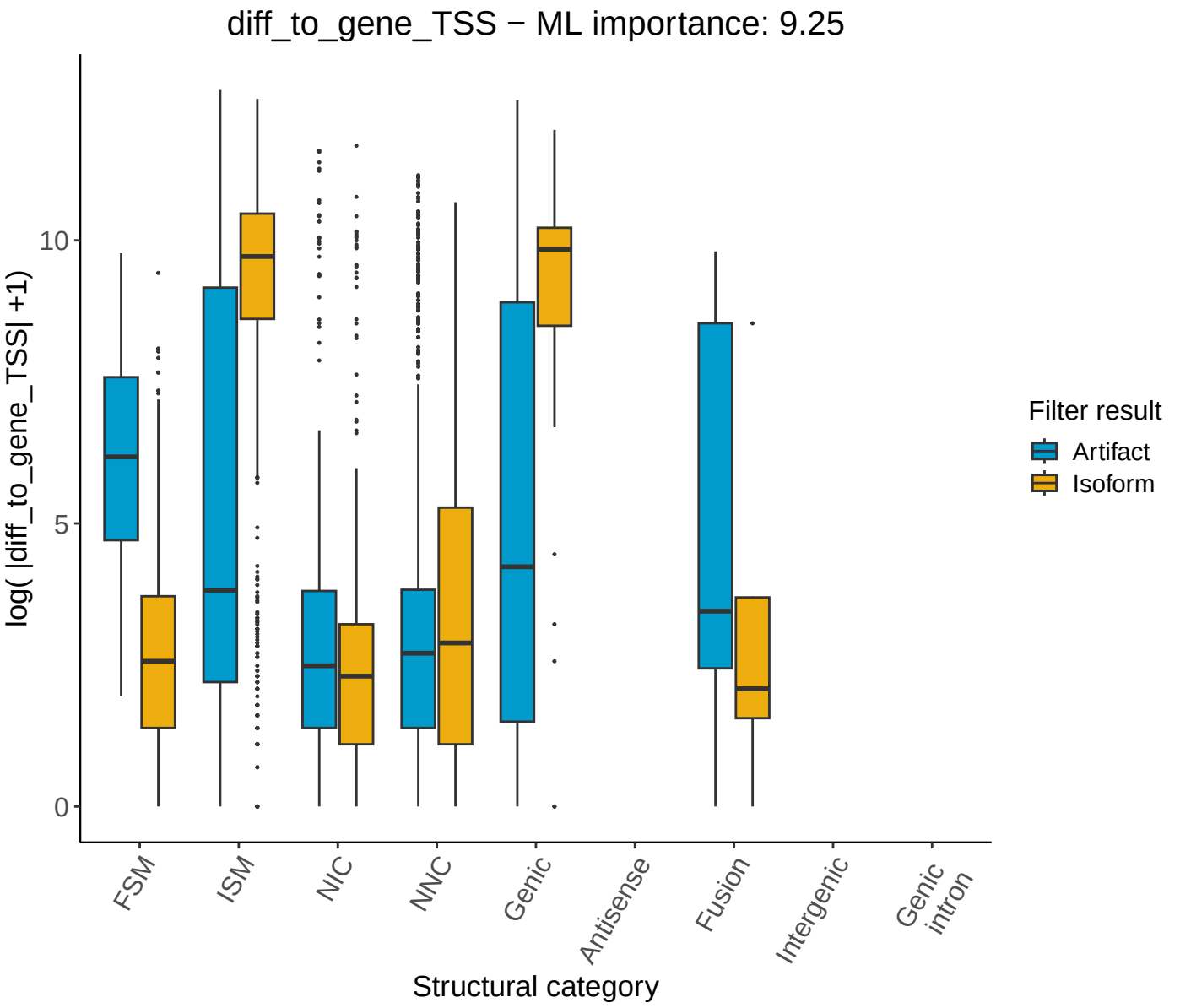

bite − ML importance: 26.35
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
506
217
718
327
373
371
116
7
4
22
4
2
2
100%
75%
50%
25%
0%
% transcripts
Isoform
Artifact
Isoform
Artifact
Isoform
Artifact
Isoform
Artifact
Artifact
Artifact
Isoform
Artifact
Artifact
Filter result
bite
FALSE
TRUE

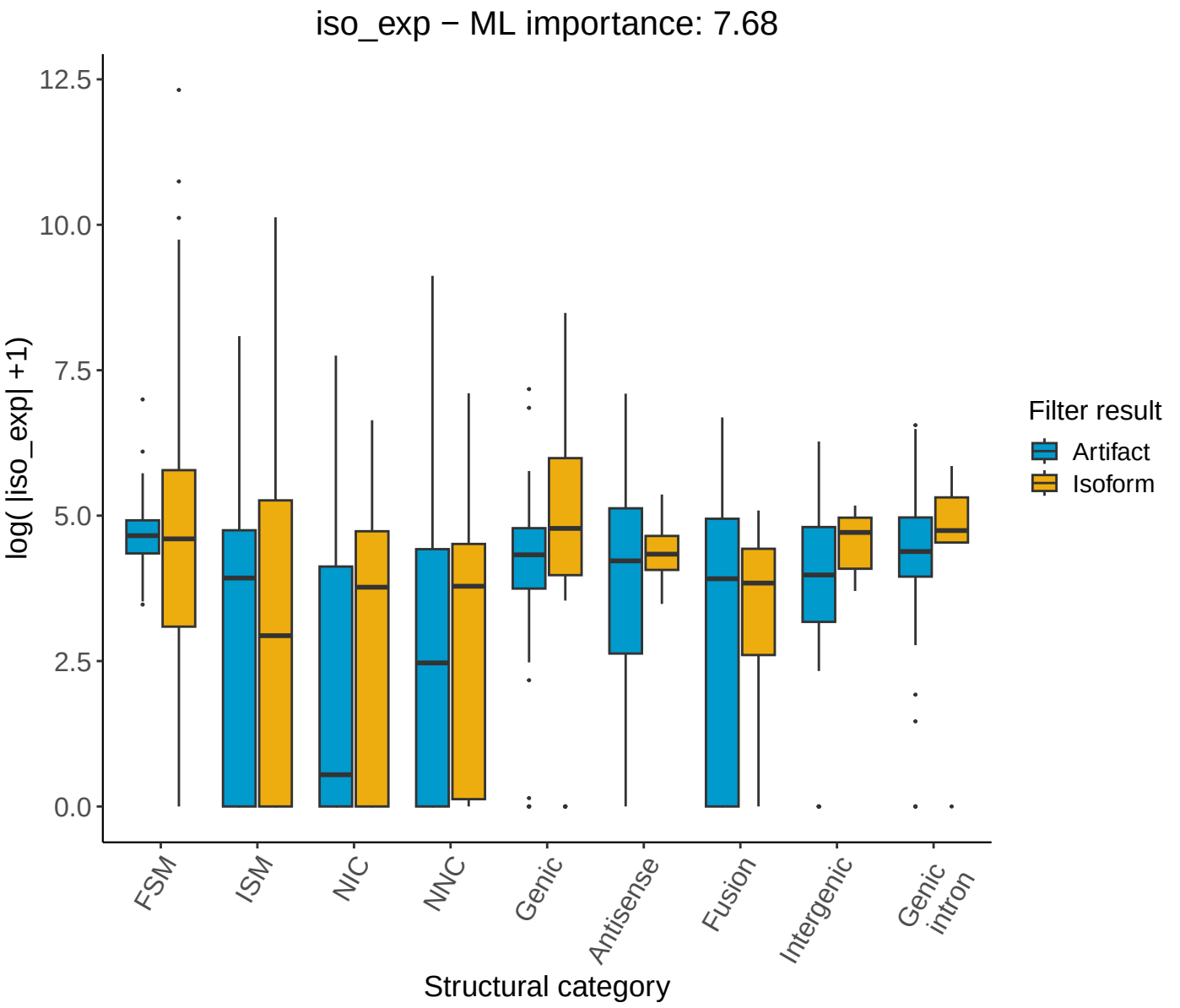

FL − ML importance: 24.39
log( |FL| +1)
7.5
5.0
2.5
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

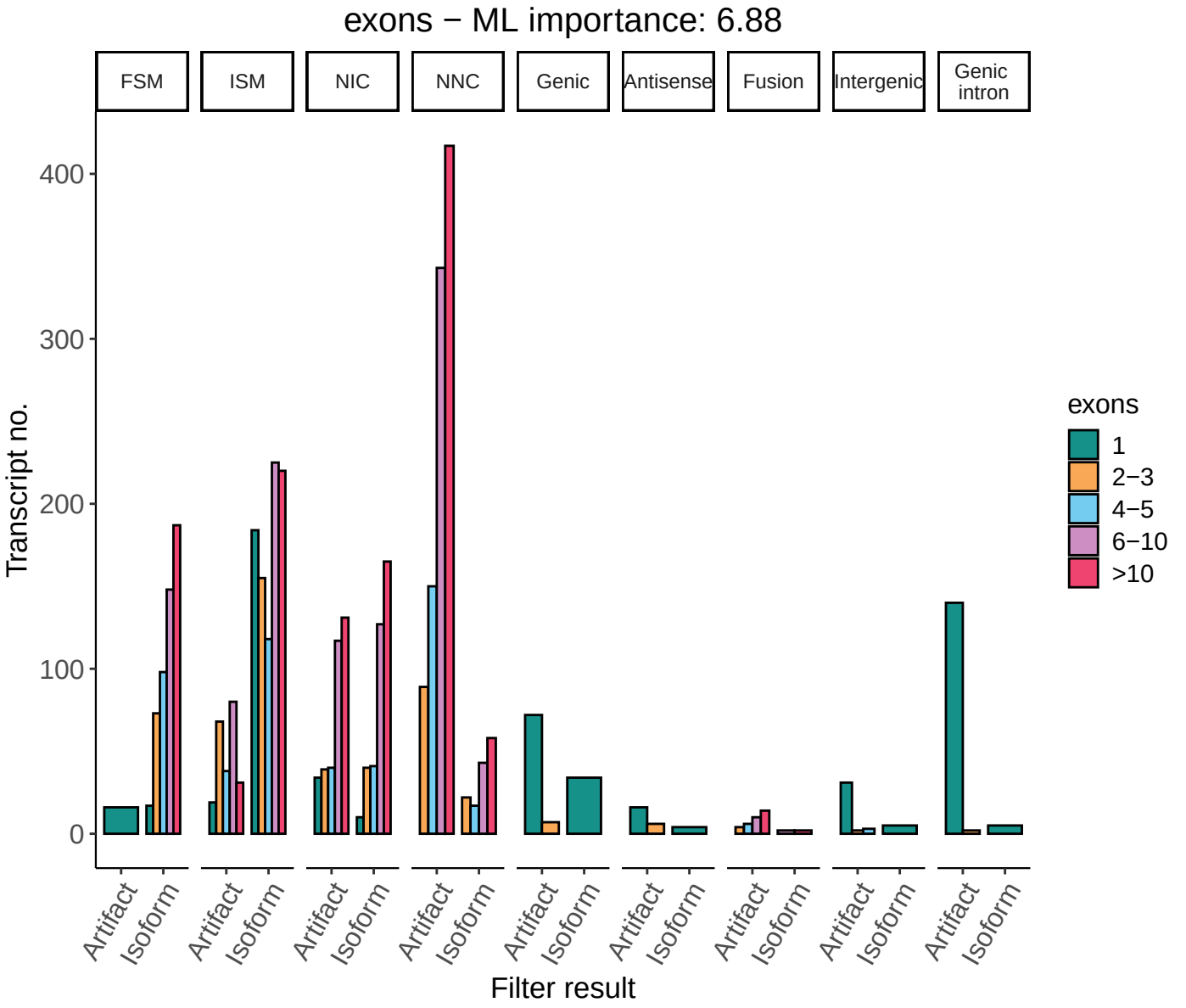

gene_exp − ML importance: 20.85
log( |gene_exp| +1)
12.5
10.0
7.5
5.0
2.5
0.0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

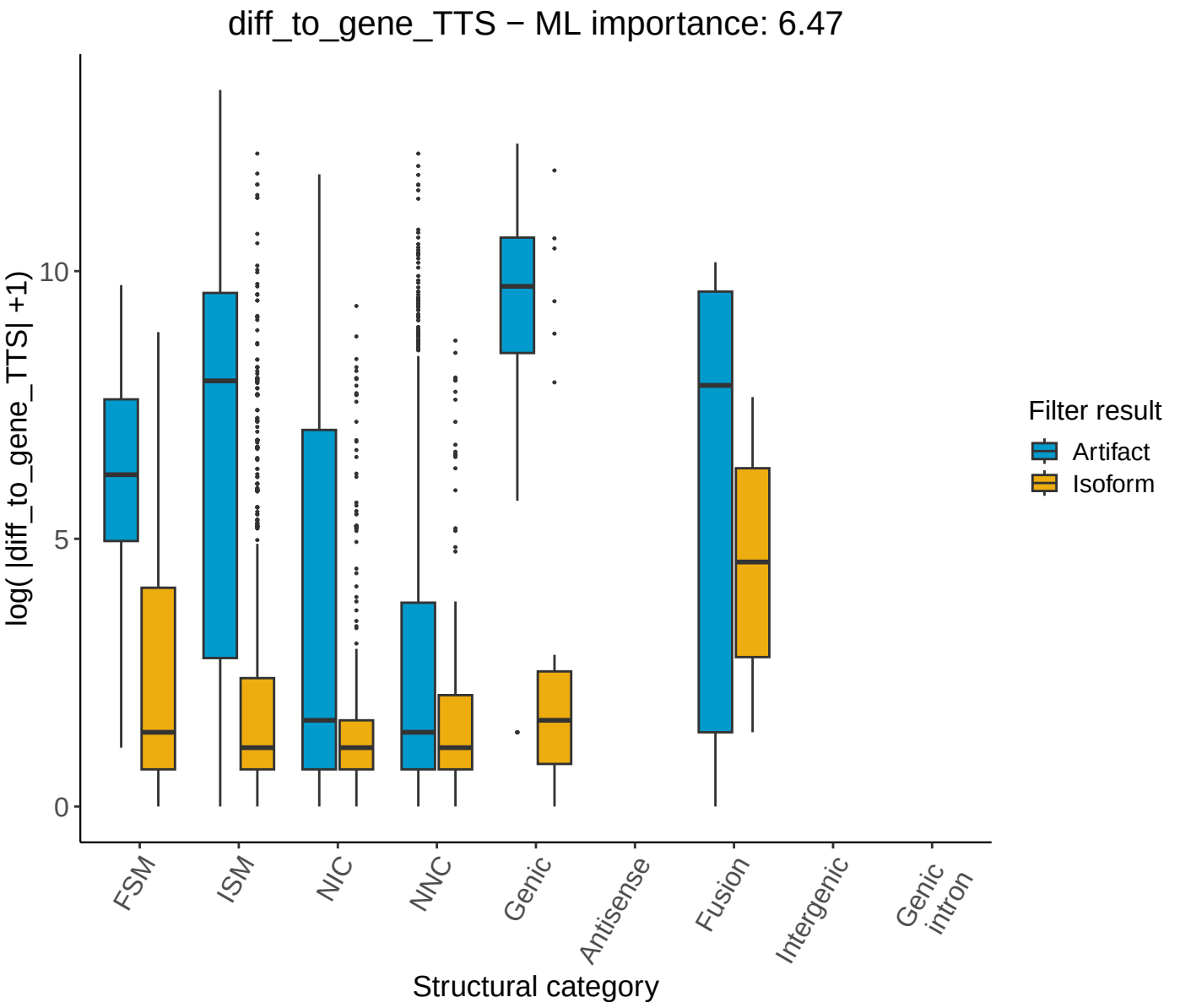

sd_cov − ML importance: 15.58
log( |sd_cov| +1)
6
4
2
0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

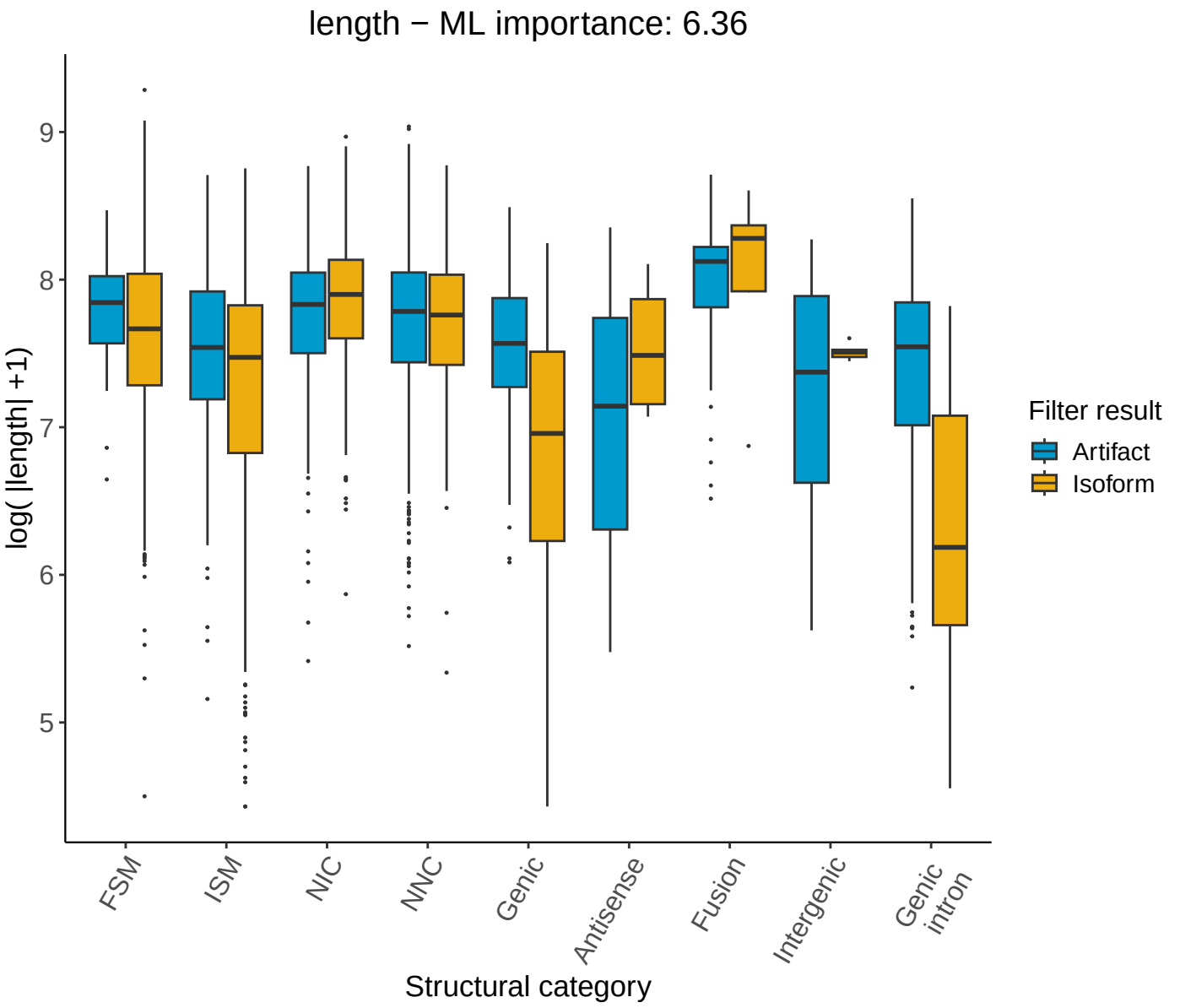

predicted_NMD − ML importance: 13.82
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
474
180
640
223
235
680
105
2
2
21
3
1
100%
75%
50%
25%
0%
% transcripts
Isoform
Artifact
Isoform
Artifact
Isoform
Artifact
Isoform
Artifact
Artifact
Artifact
Isoform
Artifact
Filter result
predicted_NMD
FALSE
TRUE

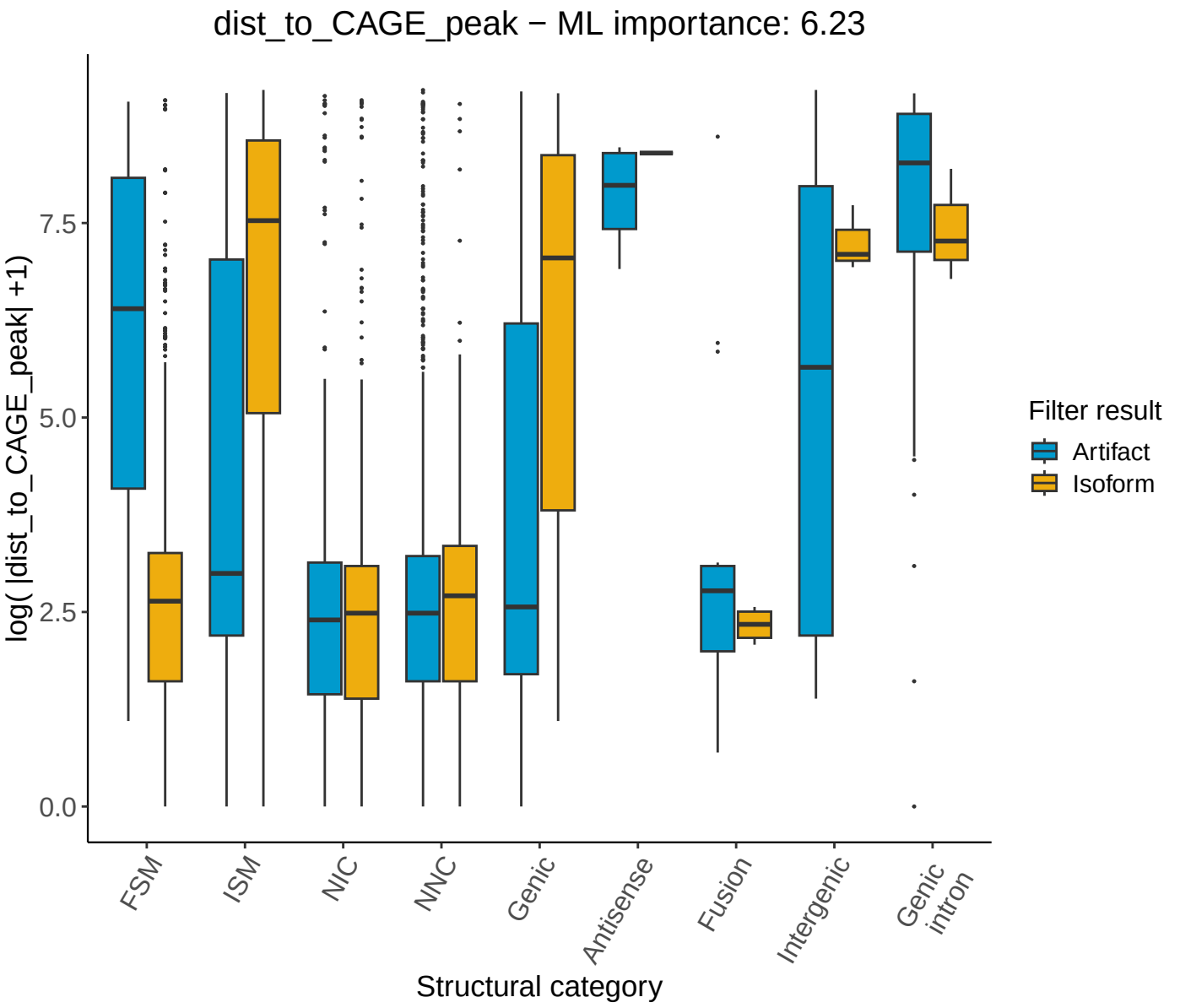

diff_to_gene_TSS − ML importance: 9.25
log( |diff_to_gene_TSS| +1)
10
5
0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

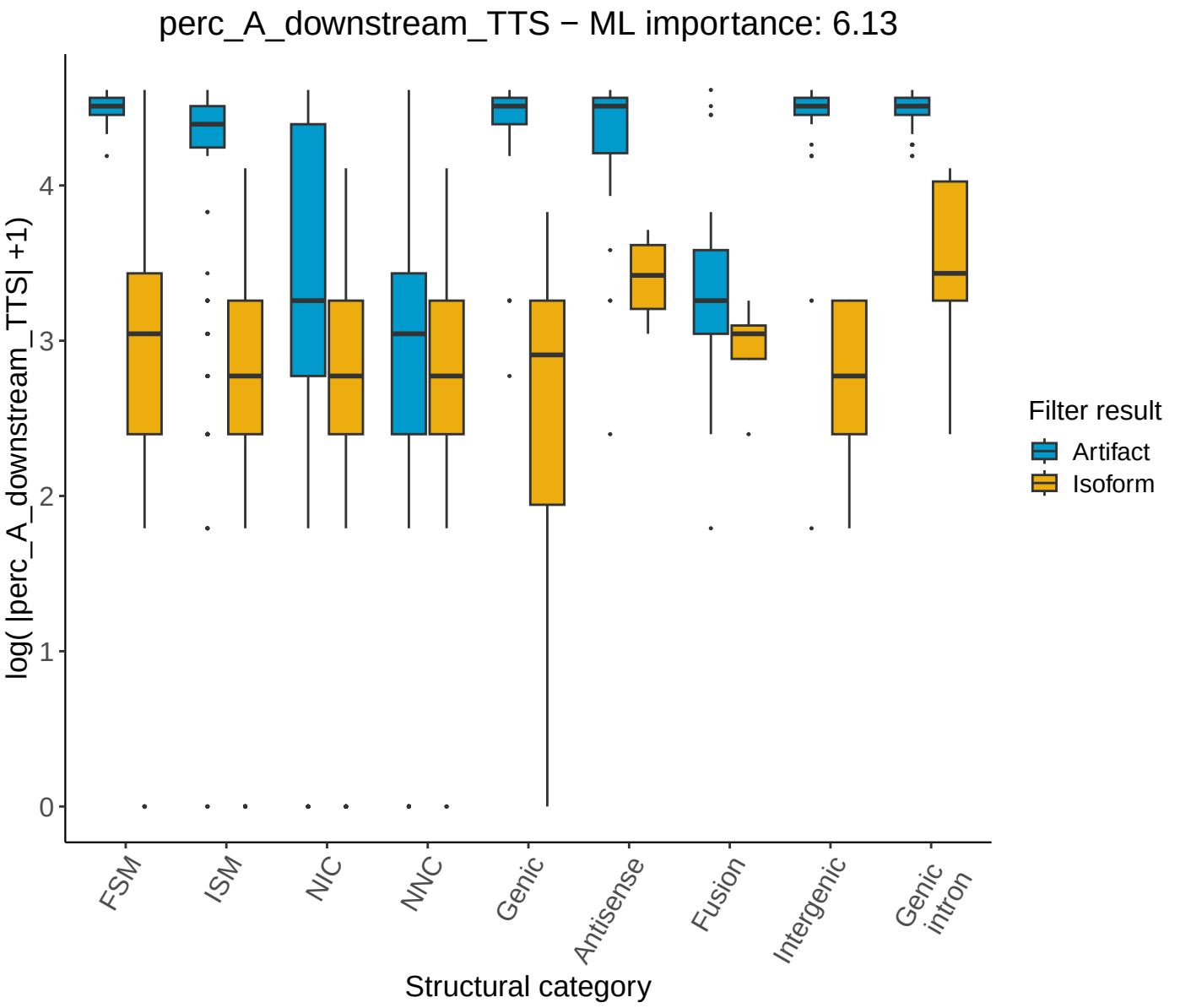

iso_exp − ML importance: 7.68
log( |iso_exp| +1)
12.5
10.0
7.5
5.0
2.5
0.0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

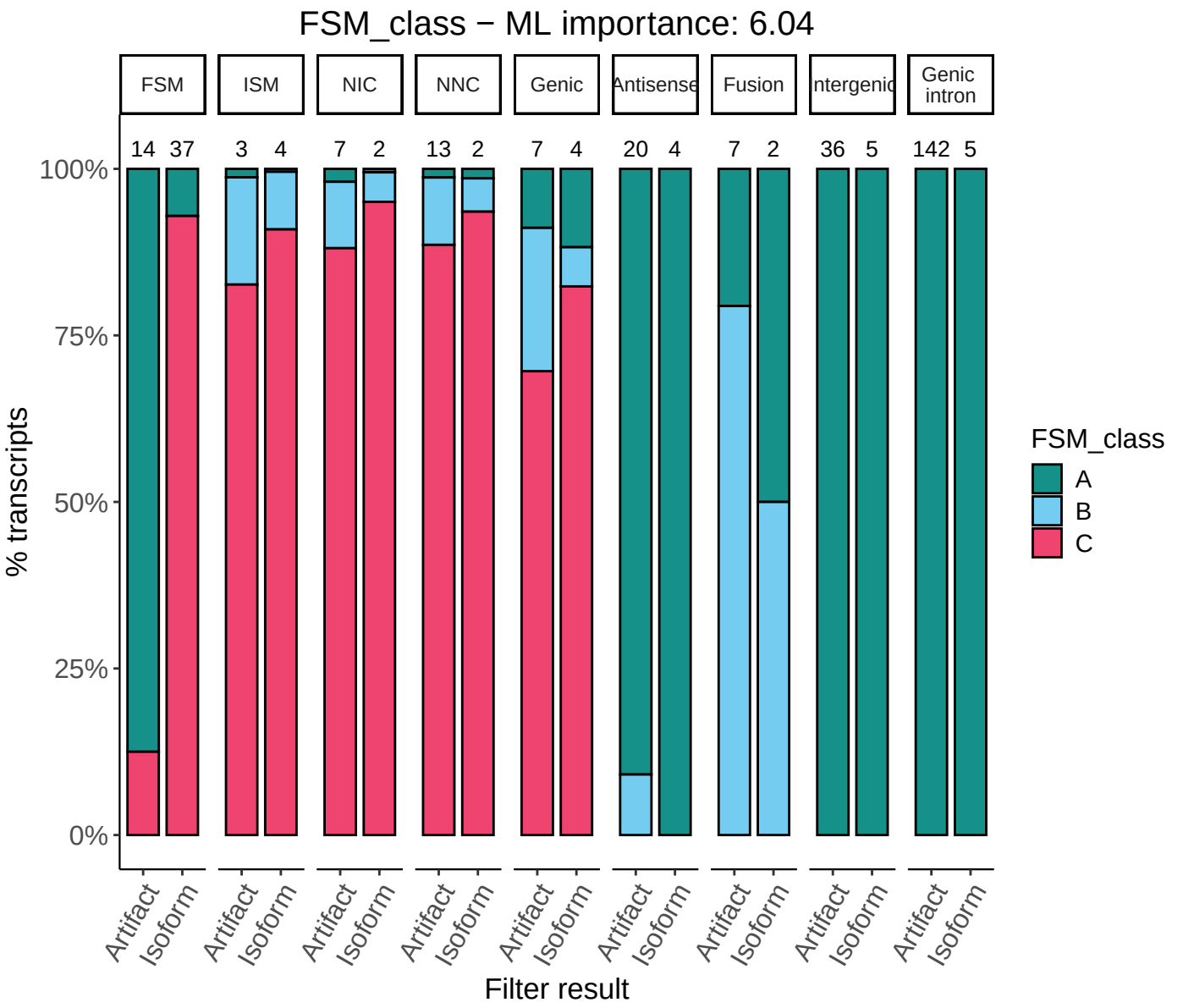

exons − ML importance: 6.88
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Transcript no.
0
100
200
300
400
Artifact
Isoform
Filter result
exons
1
2−3
4−5
6−10
>10

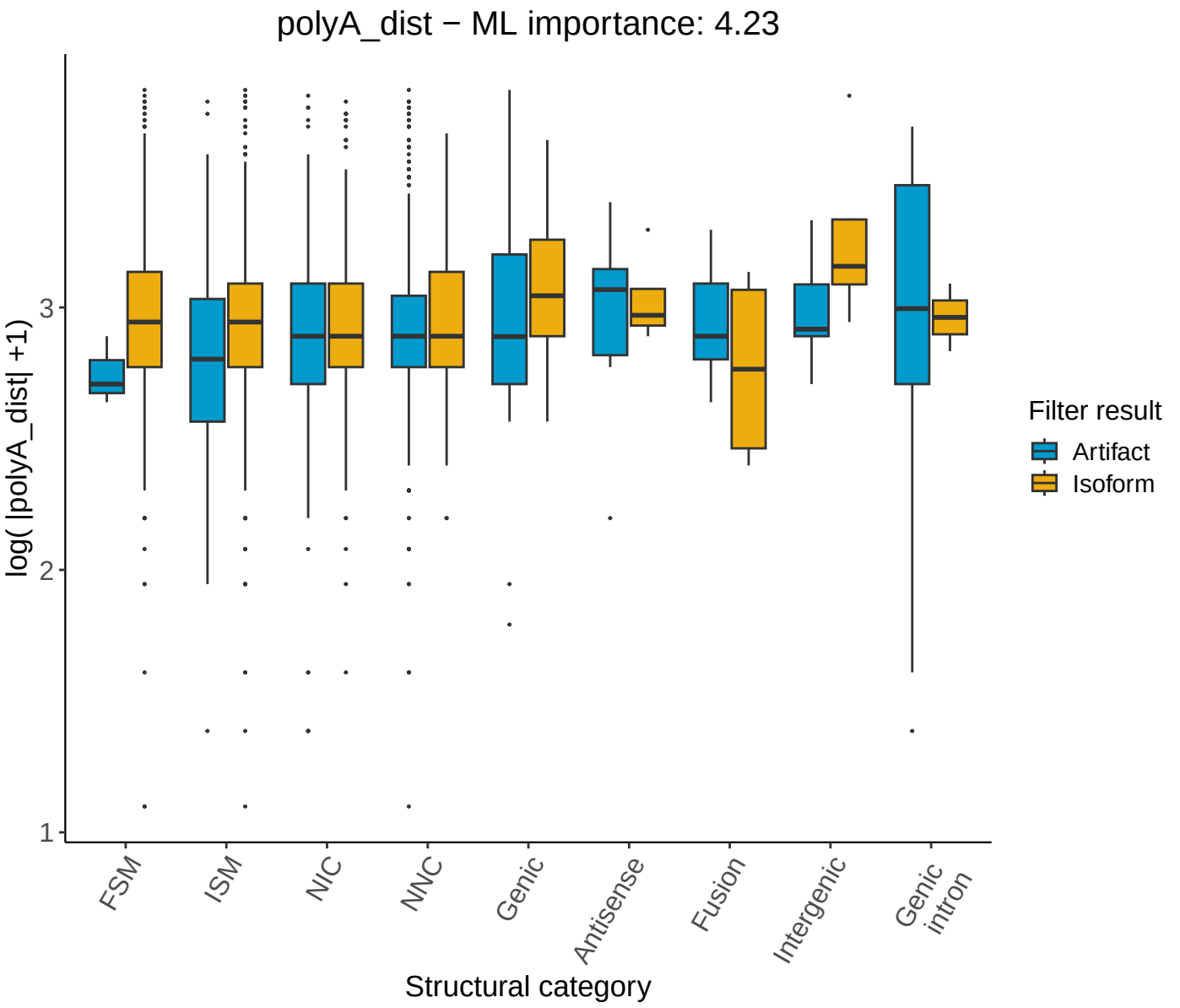

diff_to_gene_TTS − ML importance: 6.47
log( |diff_to_gene_TTS| +1)
10
5
0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

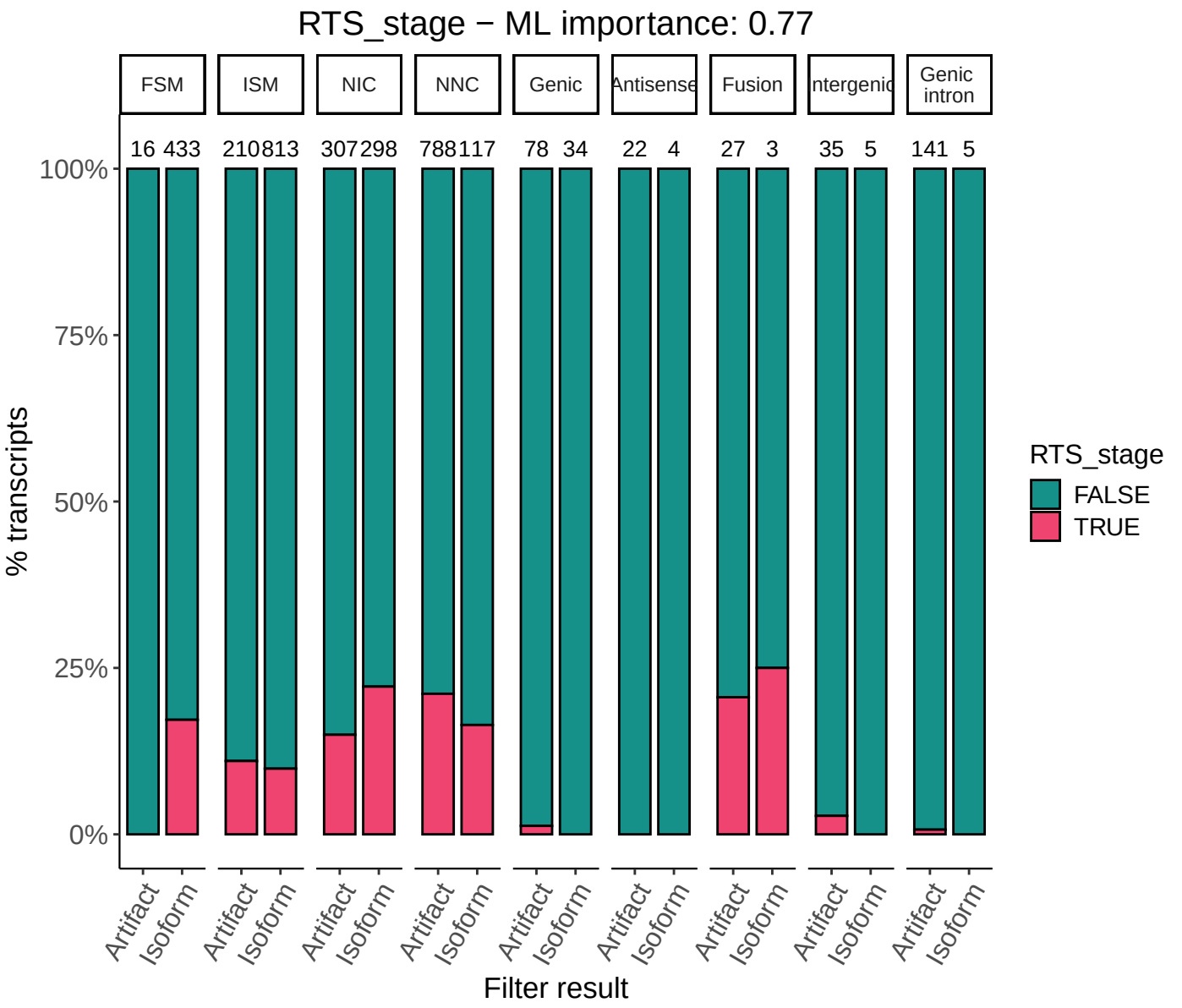

length − ML importance: 6.36
log( |length| +1)
9
8
7
6
5
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

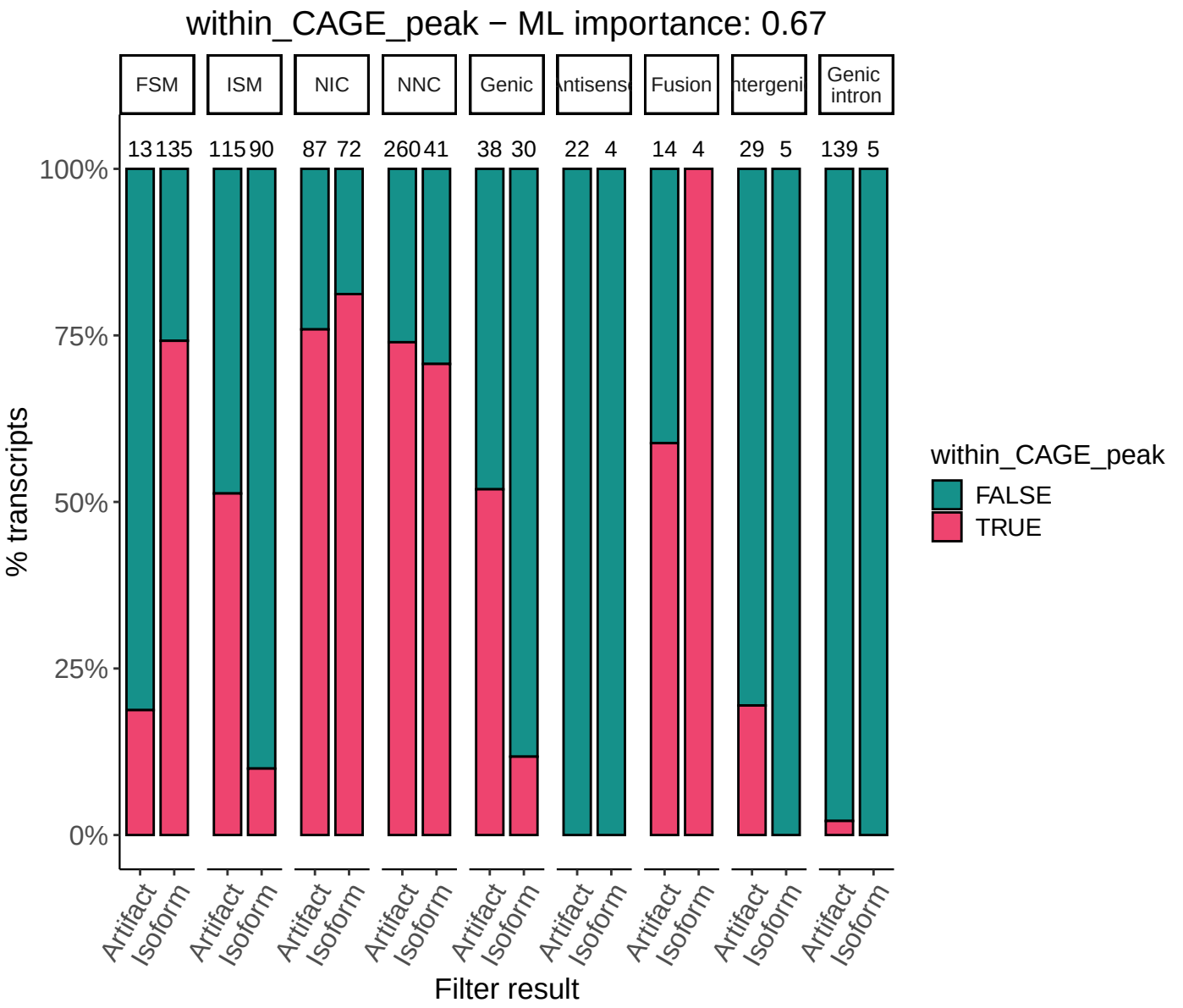

dist_to_CAGE_peak − ML importance: 6.23
log( |dist_to_CAGE_peak| +1)
7.5
5.0
2.5
0.0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

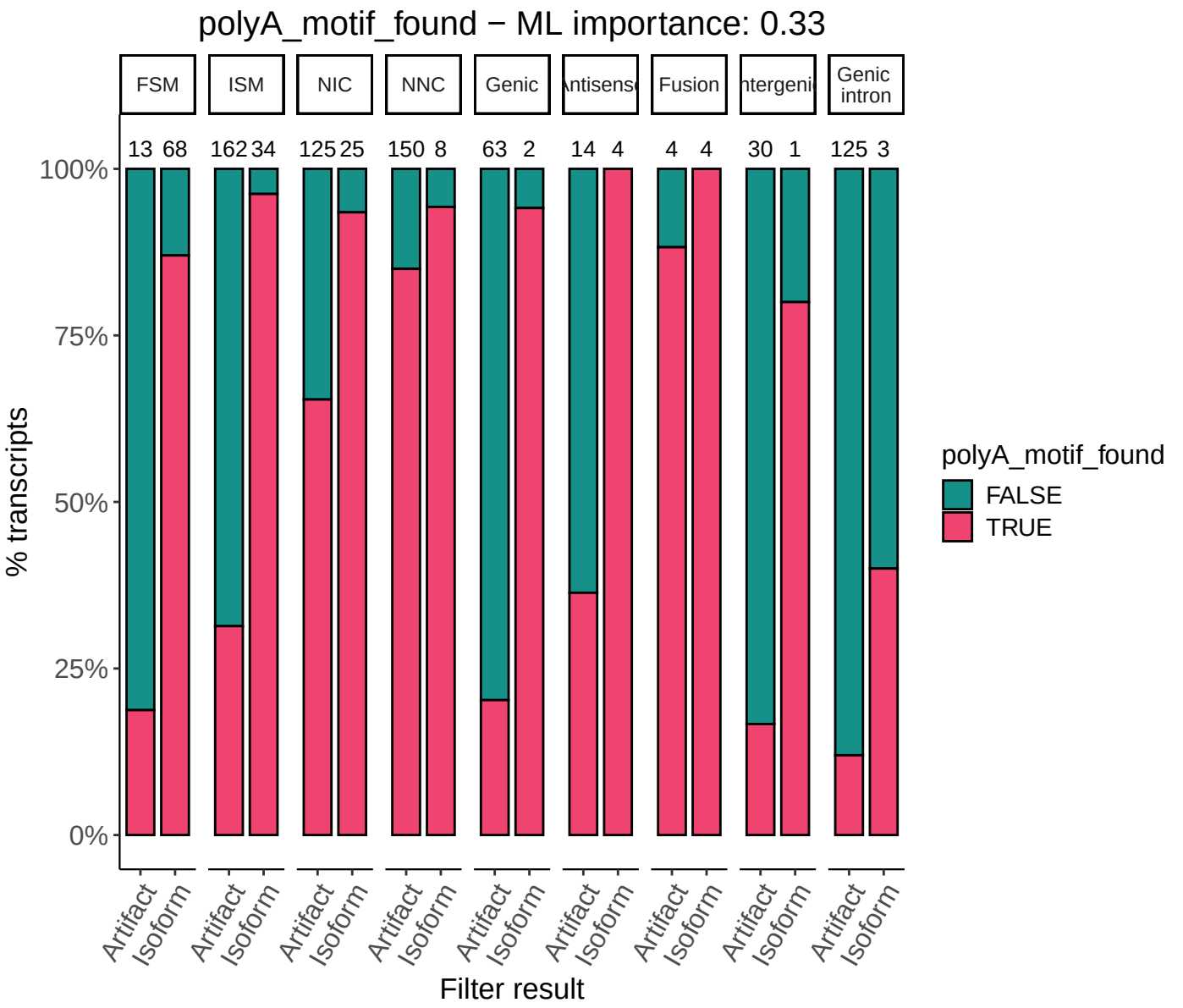

perc_A_downstream_TTS − ML importance: 6.13
log( |perc_A_downstream_TTS| +1)
0
1
2
3
4
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

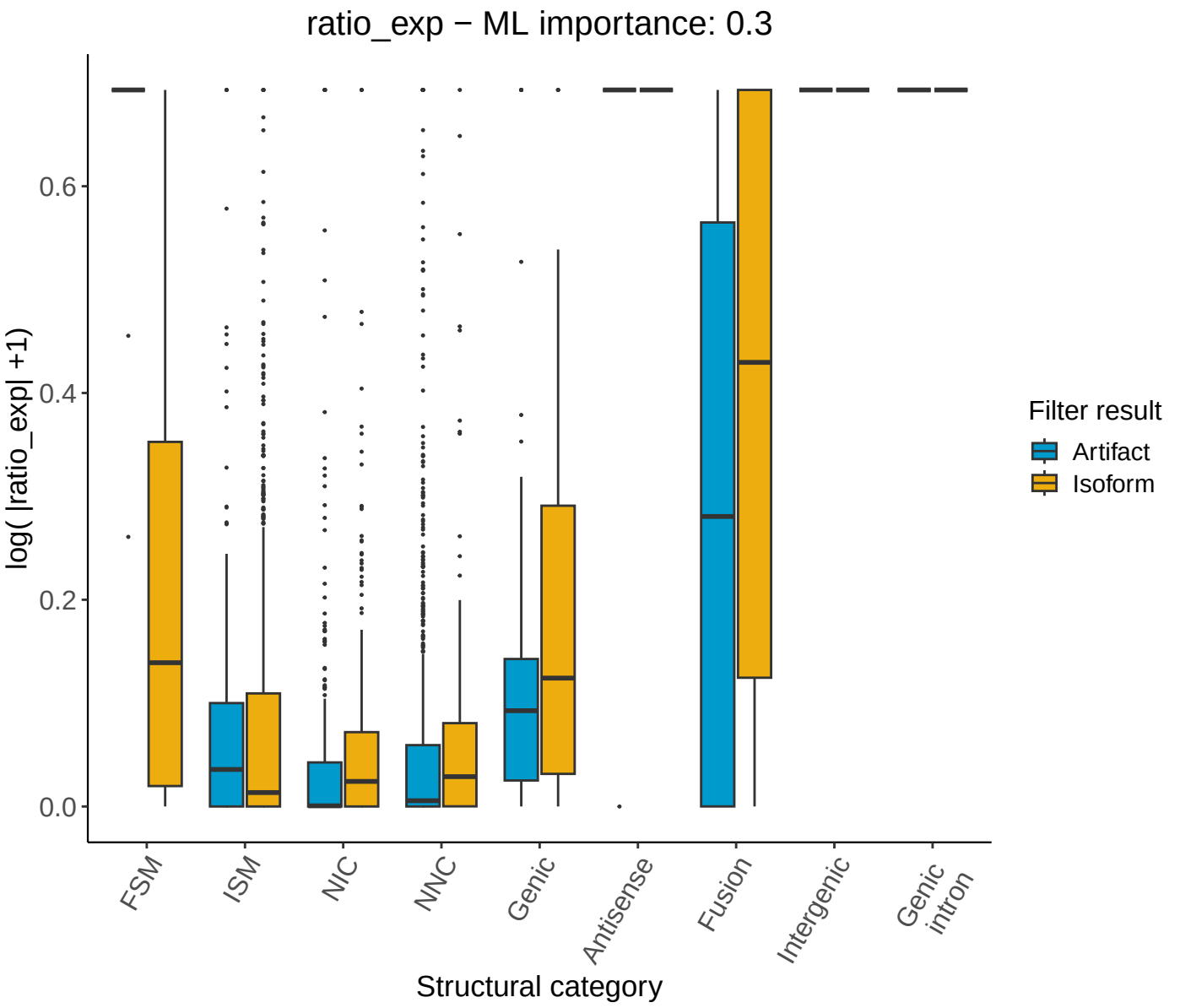

FSM_class − ML importance: 6.04
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
ntergenic
Genic intron
14 37
3 4
7 2
13 2
7 4
20 4
7 2
36 5
142 5
100%
75%
50%
25%
0%
% transcripts
Artifact
Isoform
Filter result
FSM_class
A
B
C

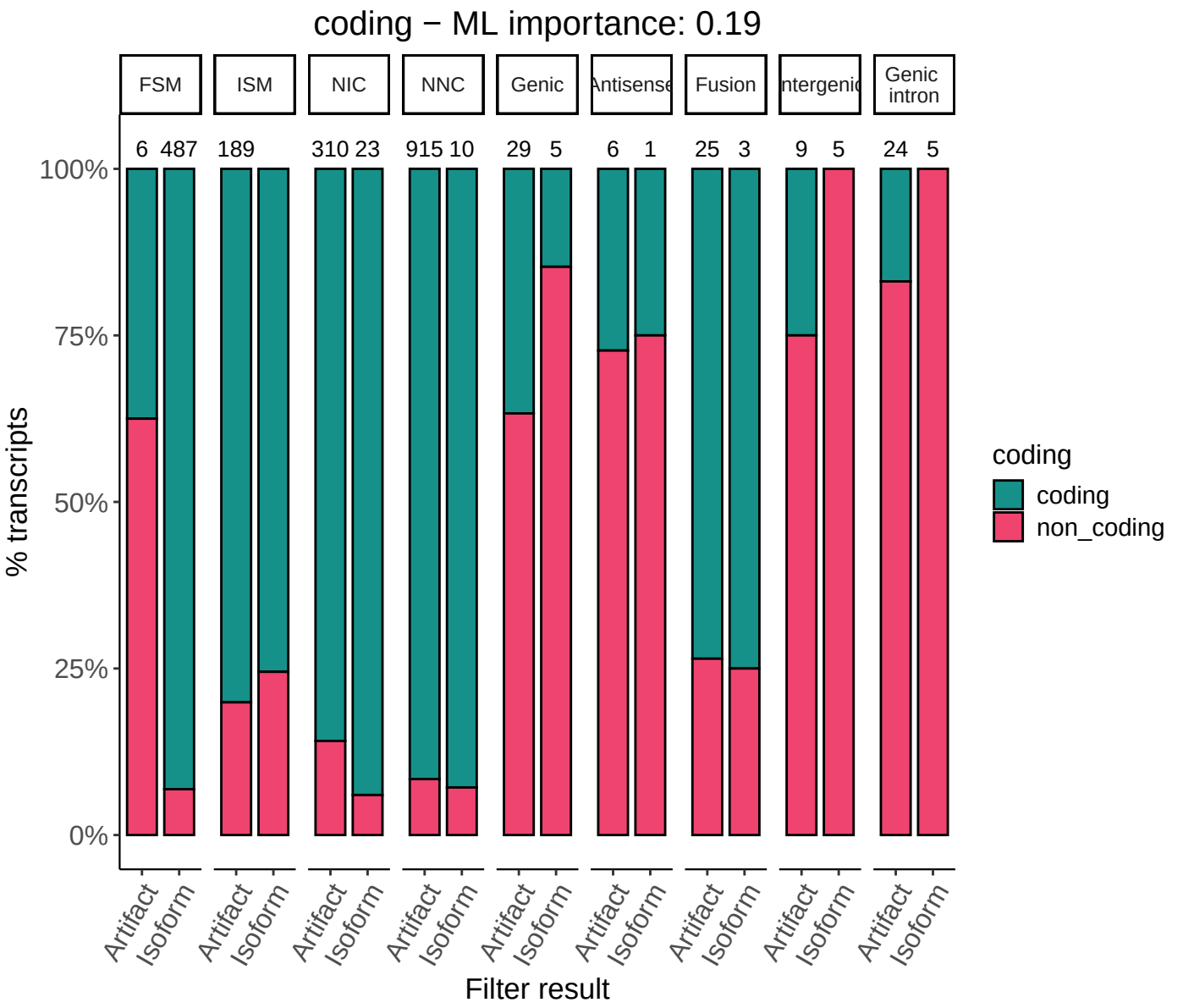

polyA_dist − ML importance: 4.23
log( |polyA_dist| +1)
3
2
1
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform


RTS_stage − ML importance: 0.77
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
ntergenic
Genic intron
16 433
210813
307298
788117
78 34
22 4
27 3
35 5
141 5
100%
75%
50%
25%
0%
% transcripts
Artifact
Isoform
Filter result
RTS_stage
FALSE
TRUE

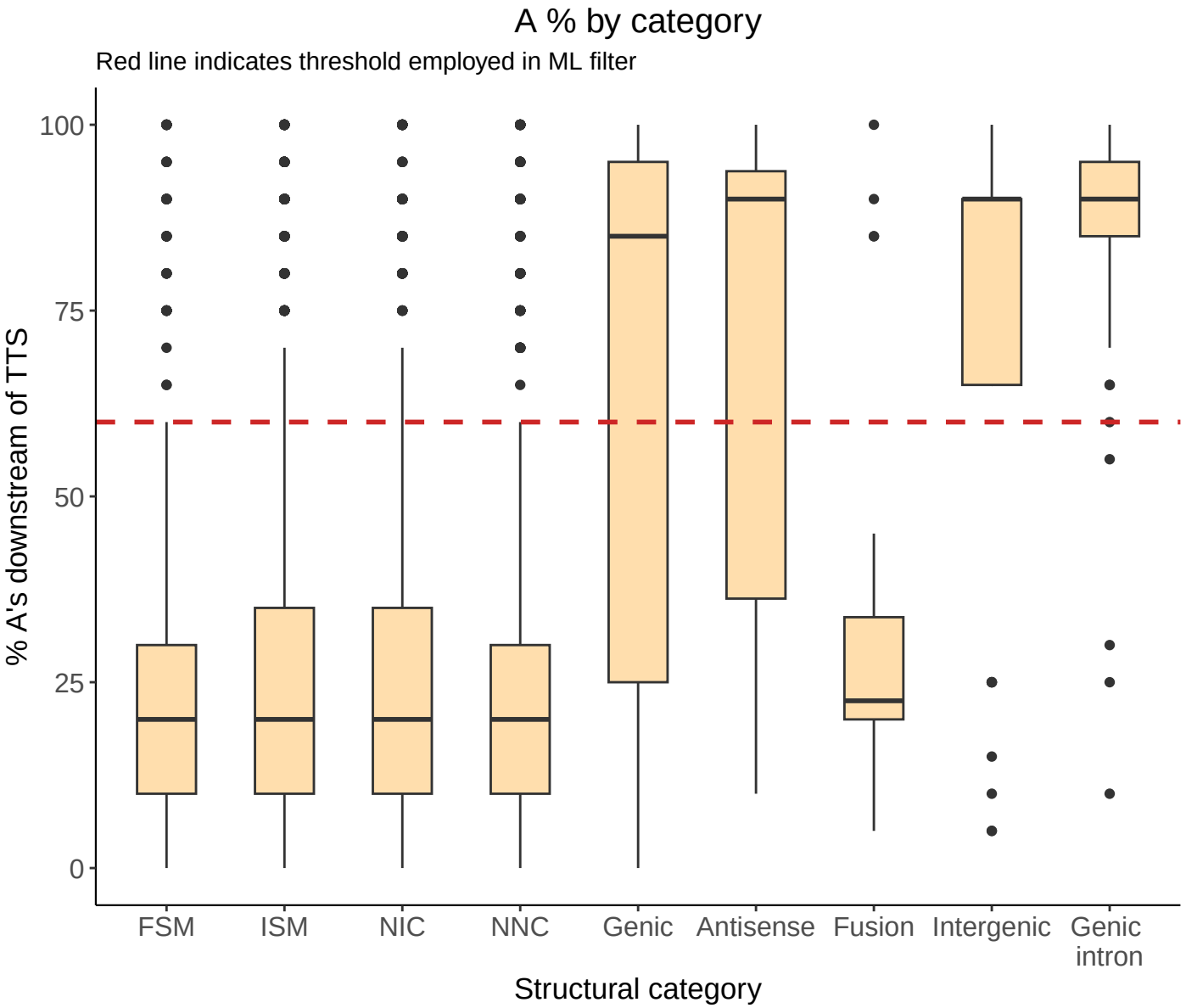

within_CAGE_peak − ML importance: 0.67
FSM
ISM
NIC
NNC
Genic
ntisens
Fusion
ntergeni
Genic intron
13 135
115 90
87 72
260 41
38 30
22 4
14 4
29 5
139 5
100%
75%
50%
25%
0%
% transcripts
Artifact
Isoform
Filter result
within_CAGE_peak
FALSE
TRUE

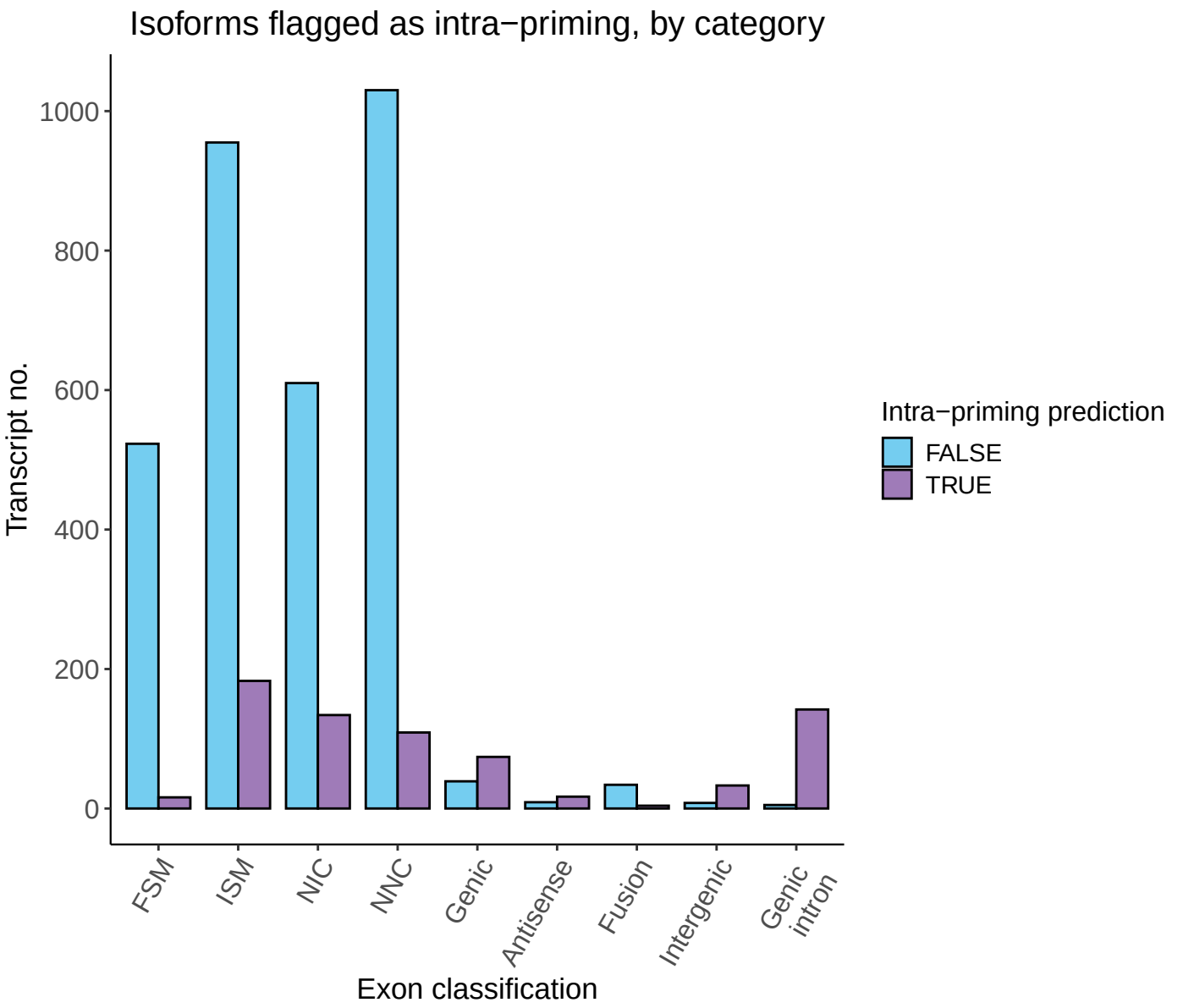

polyA_motif_found − ML importance: 0.33
FSM
ISM
NIC
NNC
Genic
ntisens
Fusion
ntergeni
Genic intron
13 68
162 34
125 25
150 8
63 2
14 4
4 4
30 1
125 3
100%
75%
50%
25%
0%
% transcripts
Artifact
Isoform
Filter result
polyA_motif_found
FALSE
TRUE

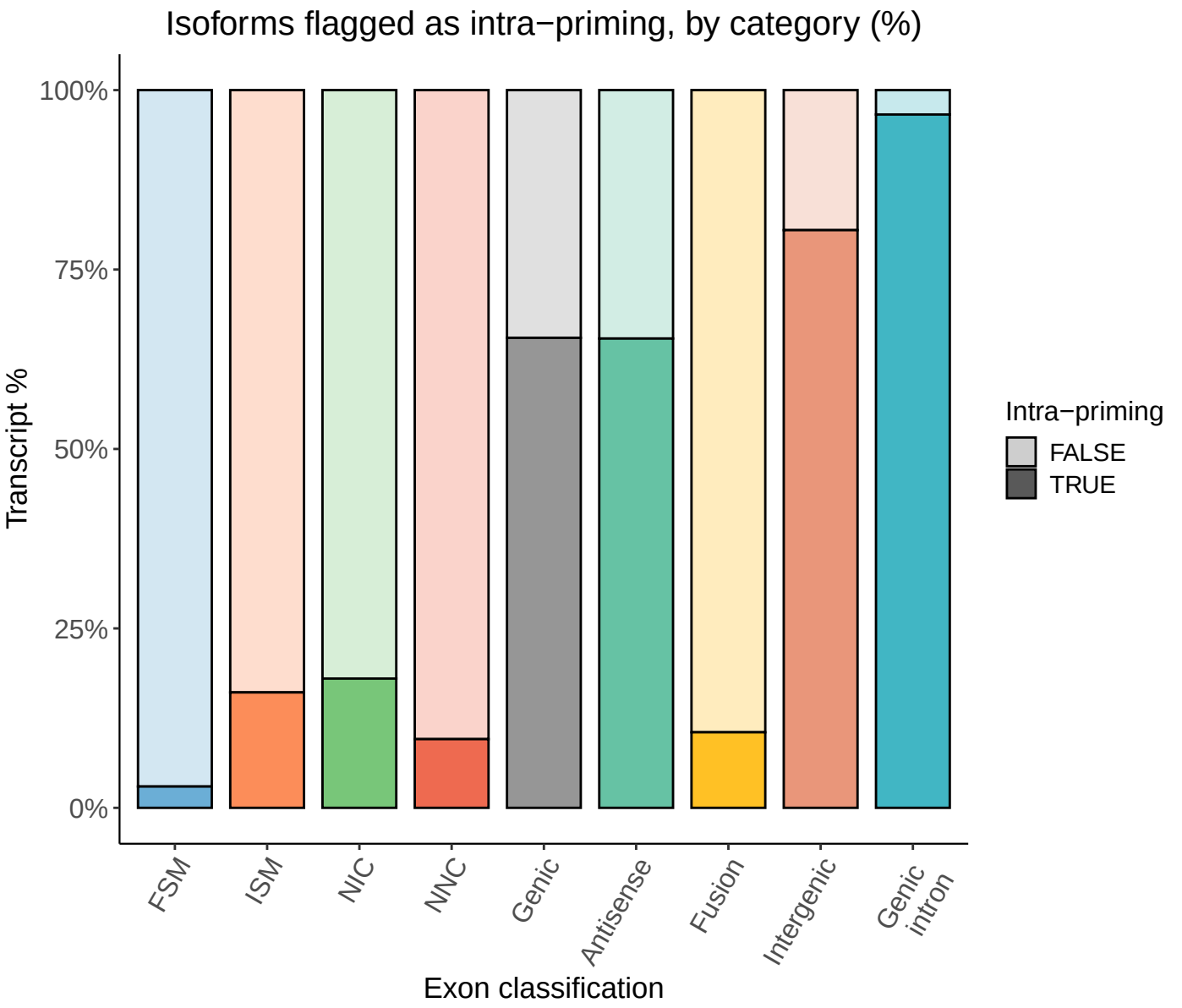

ratio_exp − ML importance: 0.3
log( |ratio_exp| +1)
0.0
0.2
0.4
0.6
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category
Filter result
Artifact
Isoform

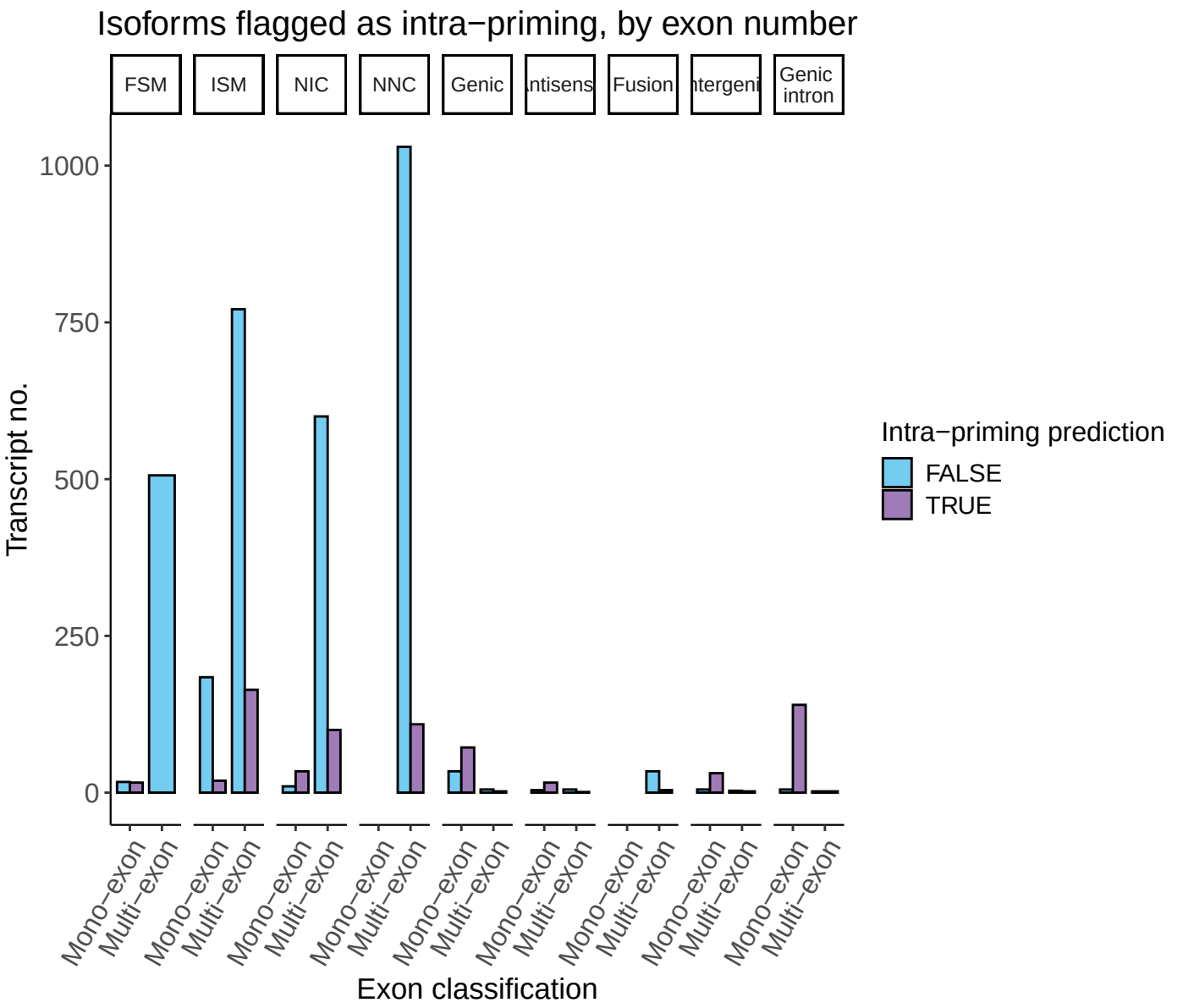

coding − ML importance: 0.19
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
6 487
189
310 23
915 10
29 5
6 1
25 3
9 5
24 5
100%
75%
50%
25%
0%
% transcripts
Artifact
Isoform
Filter result
coding
coding
non_coding

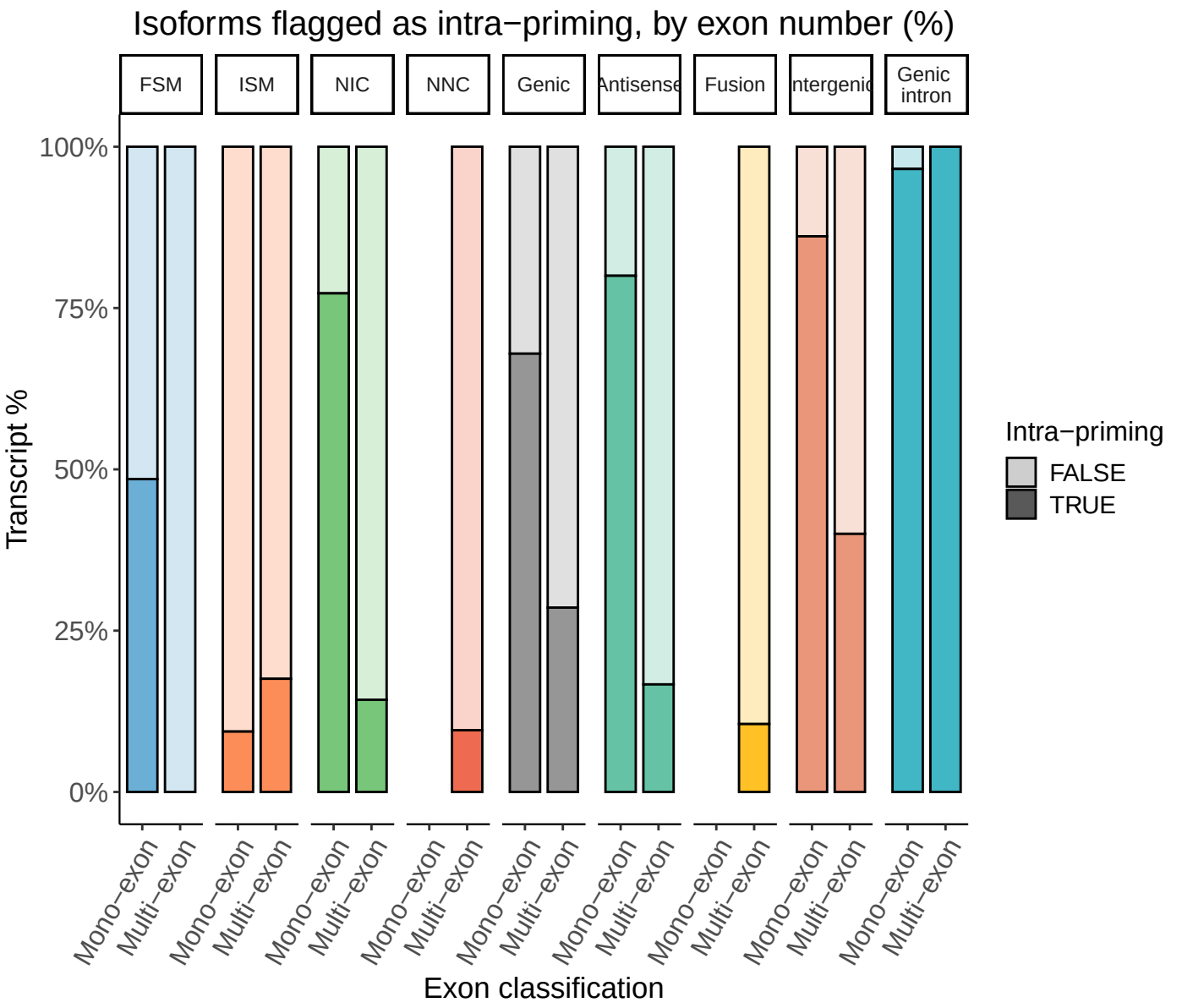
# *Intra−primming filter report*


A % by category
Red line indicates threshold employed in ML filter
% A's downstream of TTS
100
75
50
25
0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Structural category


Isoforms flagged as intra−priming, by category
Transcript no.
1000
800
600
400
200
0
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Exon classification
Intra−priming prediction
FALSE
TRUE


Isoforms flagged as intra−priming, by category (%)
100%
75%
50%
25%
0%
Transcript %
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
Intergenic
Genic intron
Exon classification
Intra−priming
FALSE
TRUE


Isoforms flagged as intra−priming, by exon number
FSM
ISM
NIC
NNC
Genic
ntisens
Fusion
ntergeni
Genic intron
1000
750
500
250
0
Transcript no.
Mono−exon
Multi−exon
Exon classification
Intra−priming prediction
FALSE
TRUE


Isoforms flagged as intra−priming, by exon number (%)
FSM
ISM
NIC
NNC
Genic
Antisense
Fusion
ntergenic
Genic intron
100%
75%
50%
25%
0%
Transcript %
Mono−exon
Multi−exon
Exon classification
Intra−priming
FALSE
TRUE