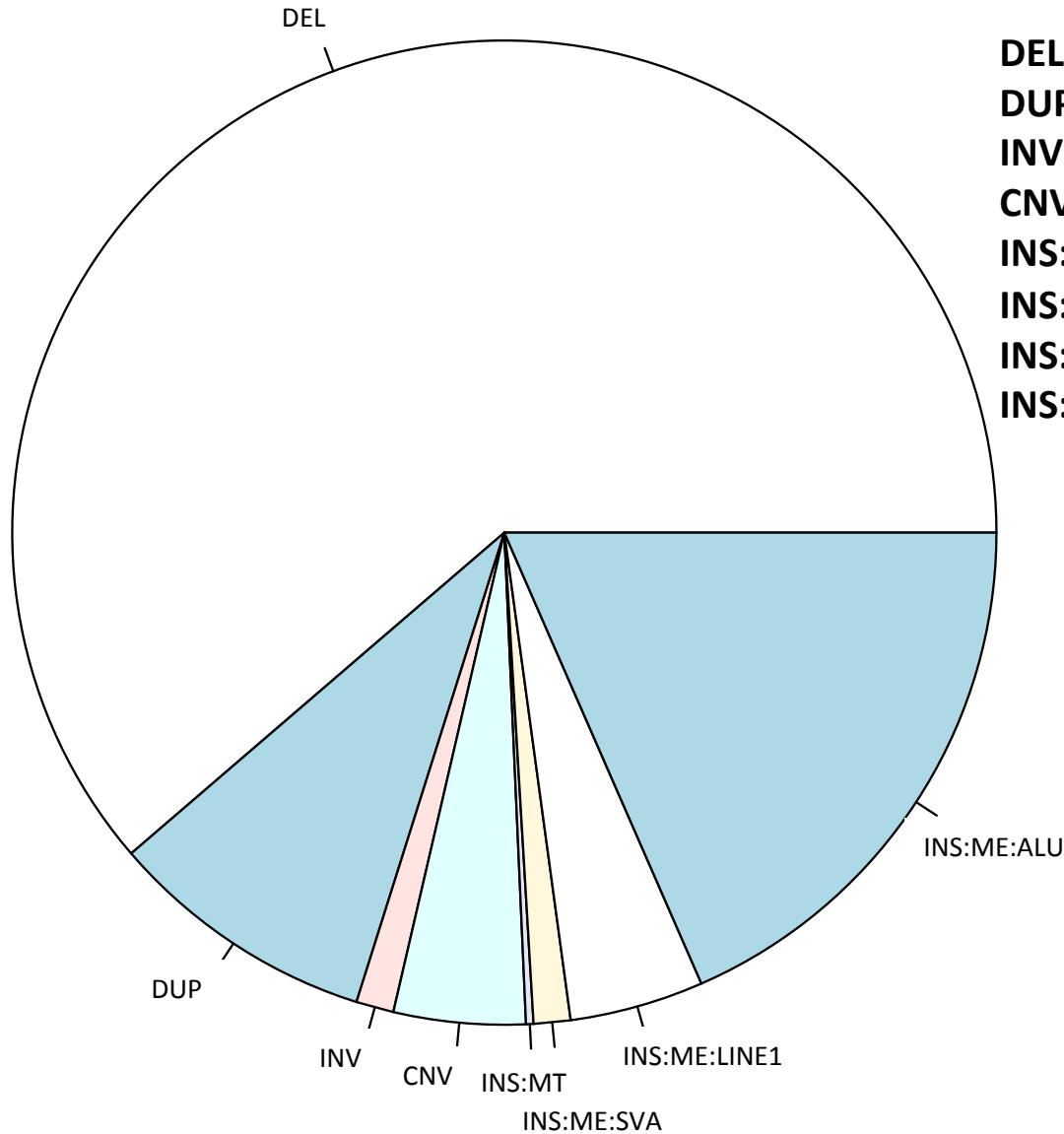


# Phase 3 SV overlap with functional regions

Yan Zhang

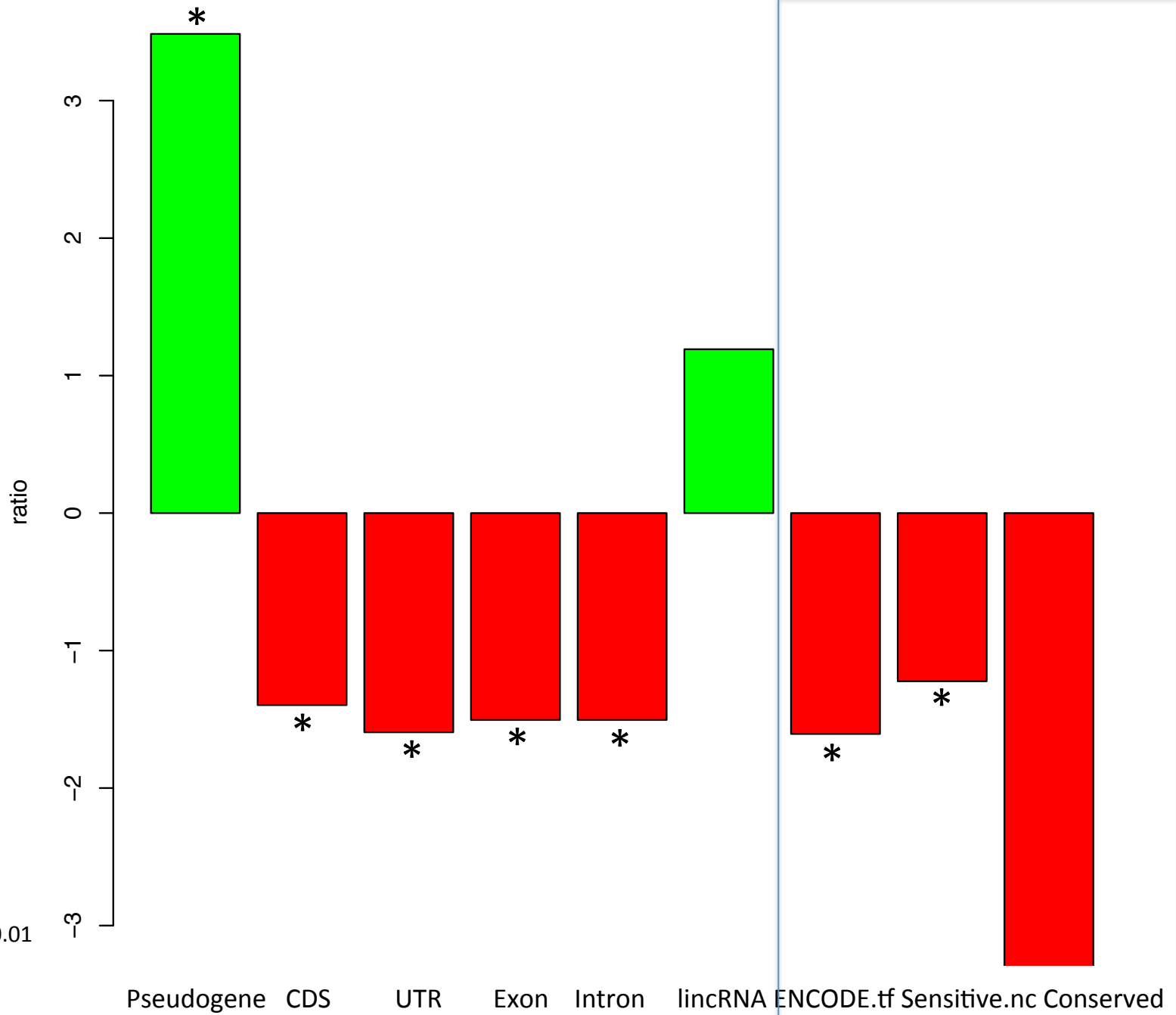
9/17/2014

# Different SV types



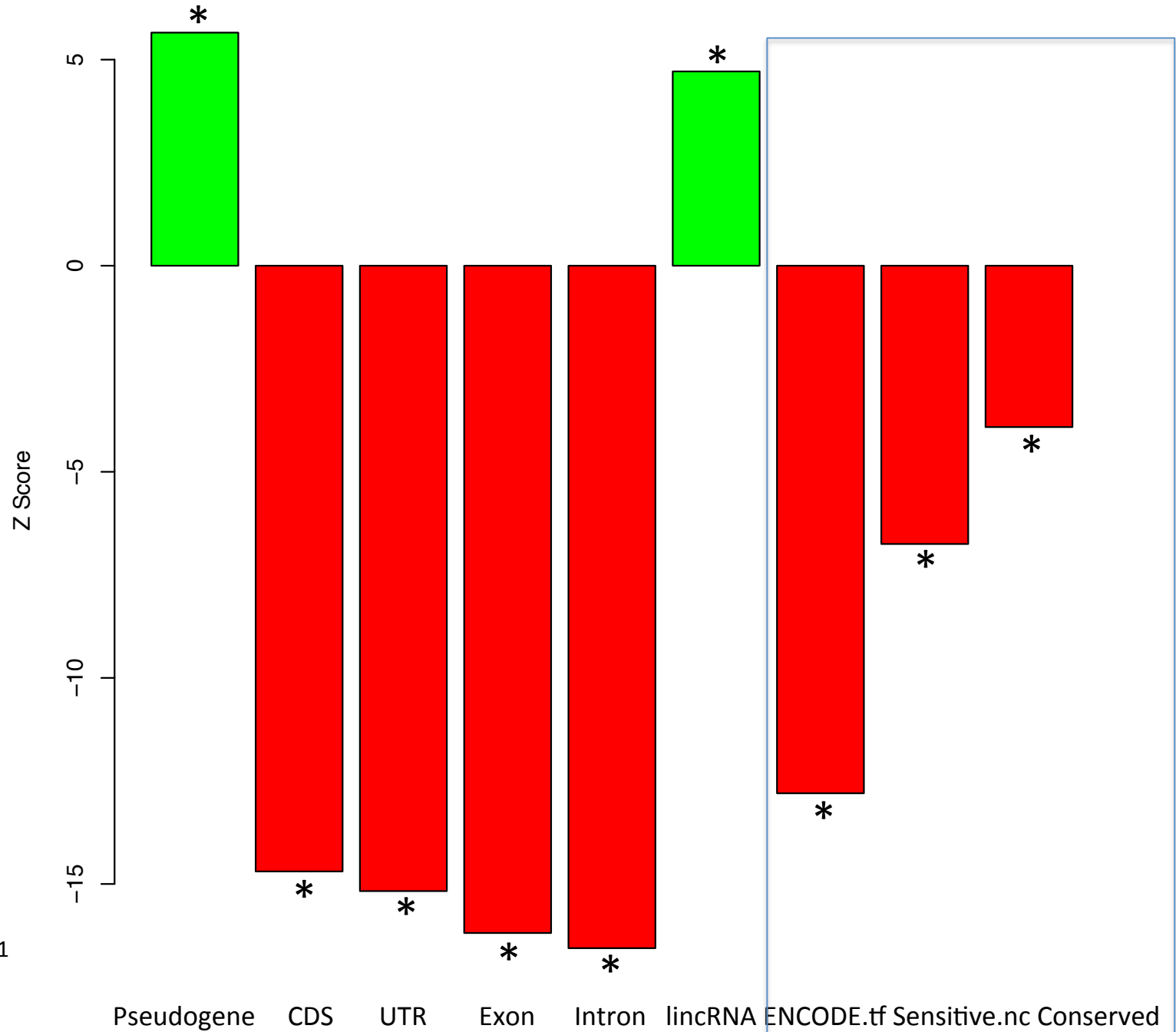
- DEL** Deletion
- DUP** Duplication
- INV** Inversion
- CNV** Copy number polymorphism
- INS:MT** Nuclear Mitochondrial Insertion
- INS:ME:SVA** Insertion of SVA element
- INS:ME:LINE1** Insertion of LINE1 element
- INS:ME:ALU** Insertion of ALU element

# CNV overlap with genomic elements



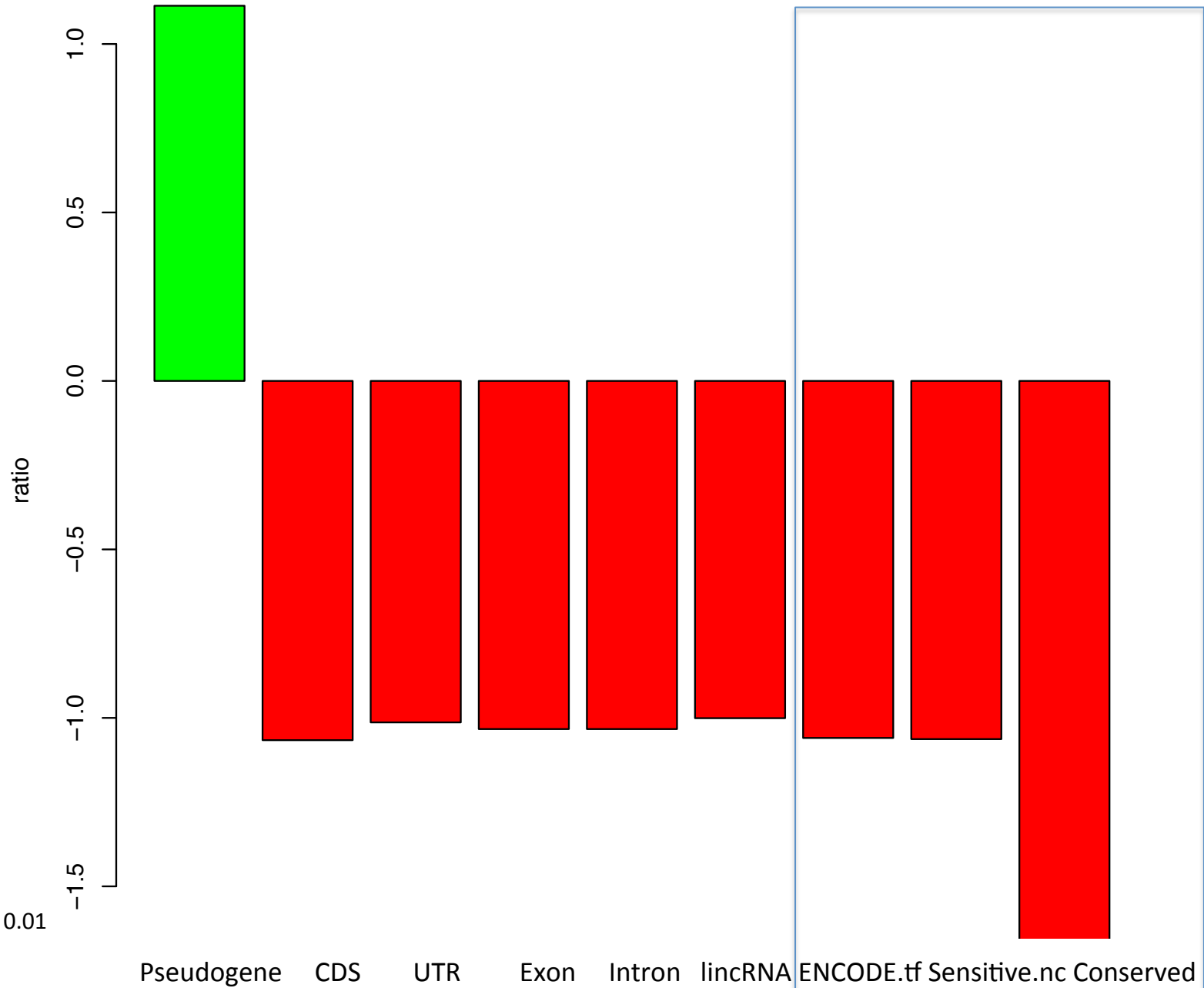
\* pvalue < 0.01

# DEL overlap with genomic elements



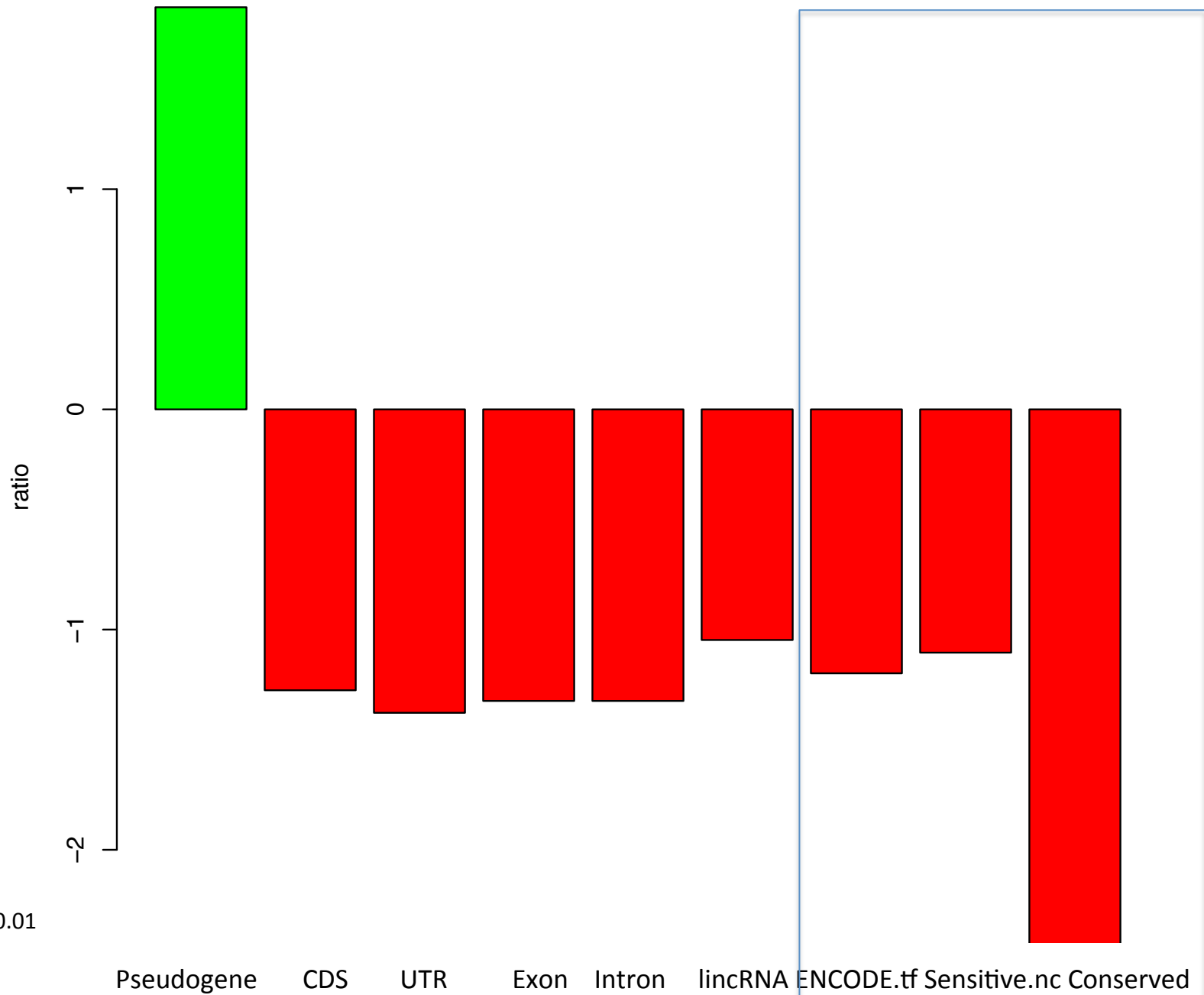
\* pvalue < 0.01

# DUP overlap with genomic elements



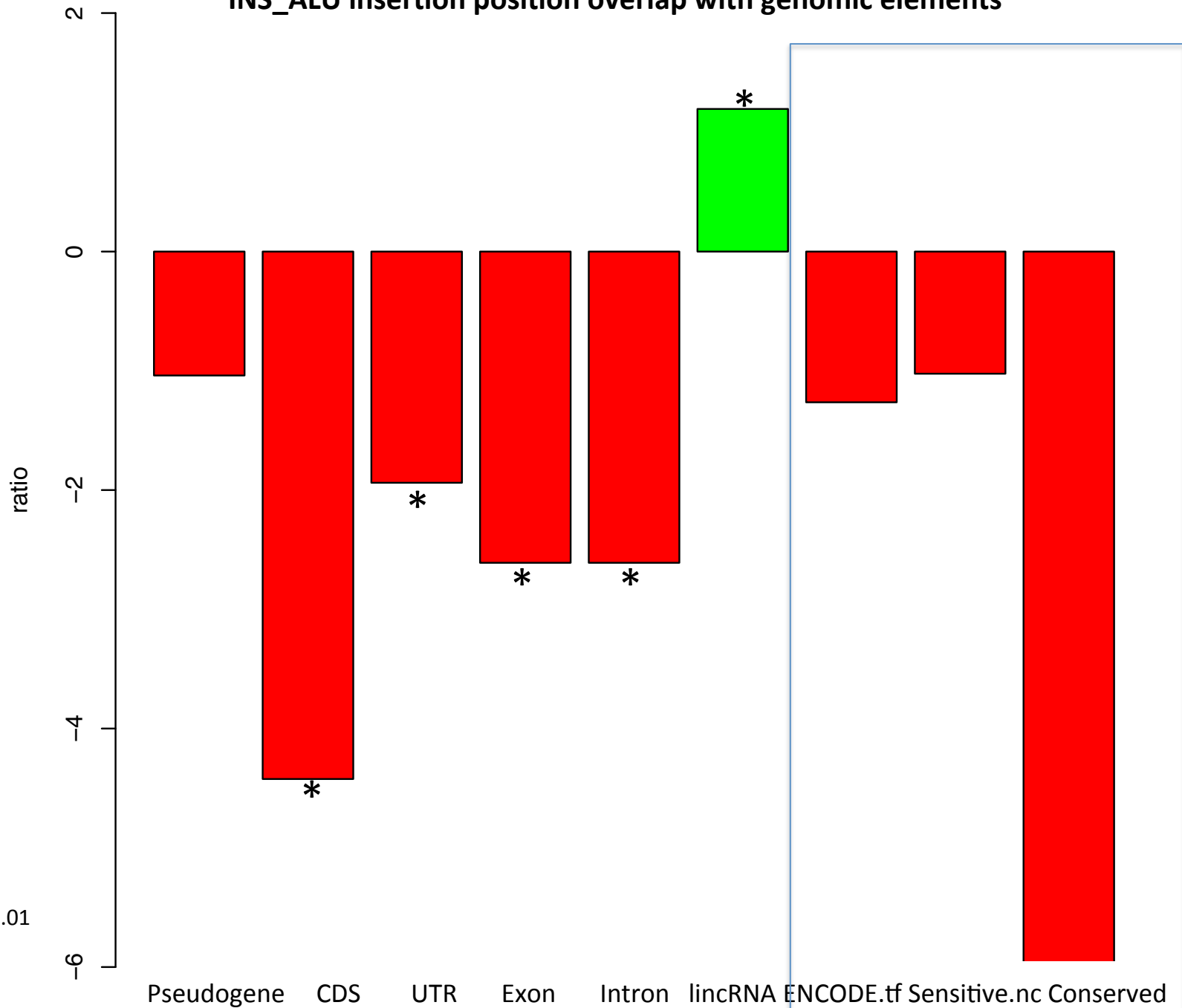
\* pvalue < 0.01

# INV overlap with genomic elements

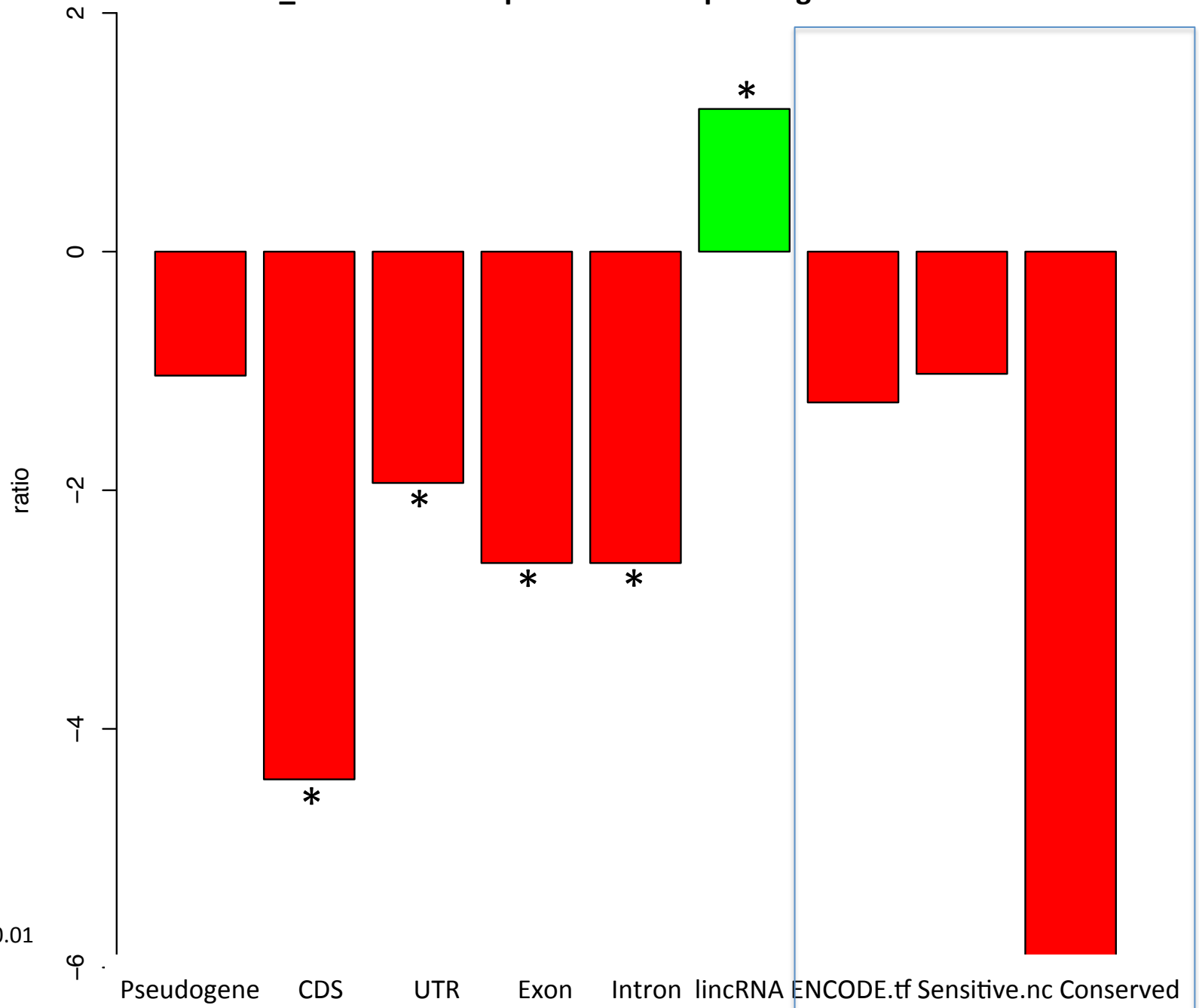


\* pvalue < 0.01

# INS\_ALU insertion position overlap with genomic elements



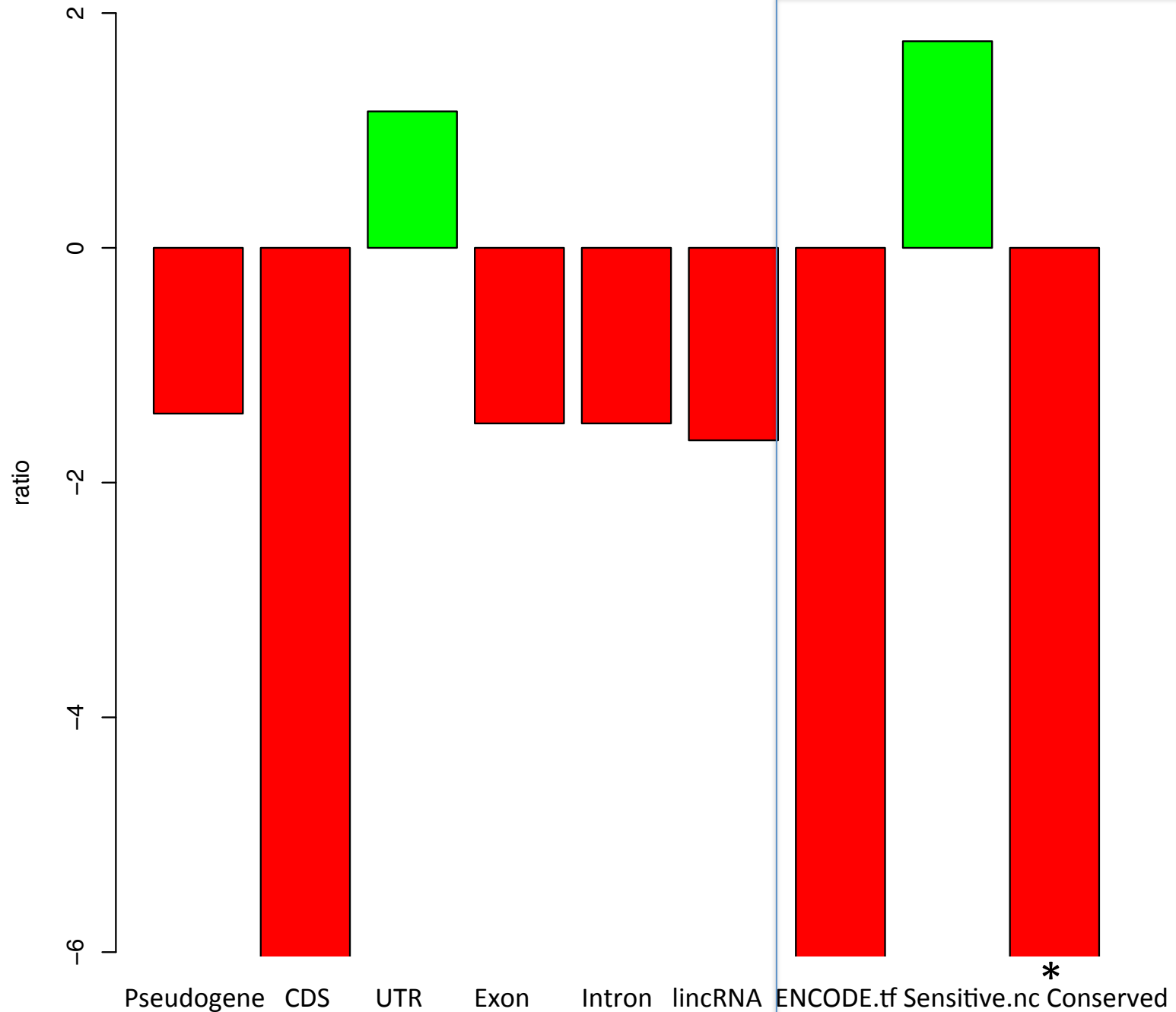
# INS\_LINE1 insertion position overlap with genomic elements



\* pvalue < 0.01



# INS\_MT insertion position overlap with genomic elements



\* pvalue < 0.01

\*

# INS\_SVA insertion position overlap with genomic elements

