# Phase 3 SV overlap with functional regions

Yan Zhang
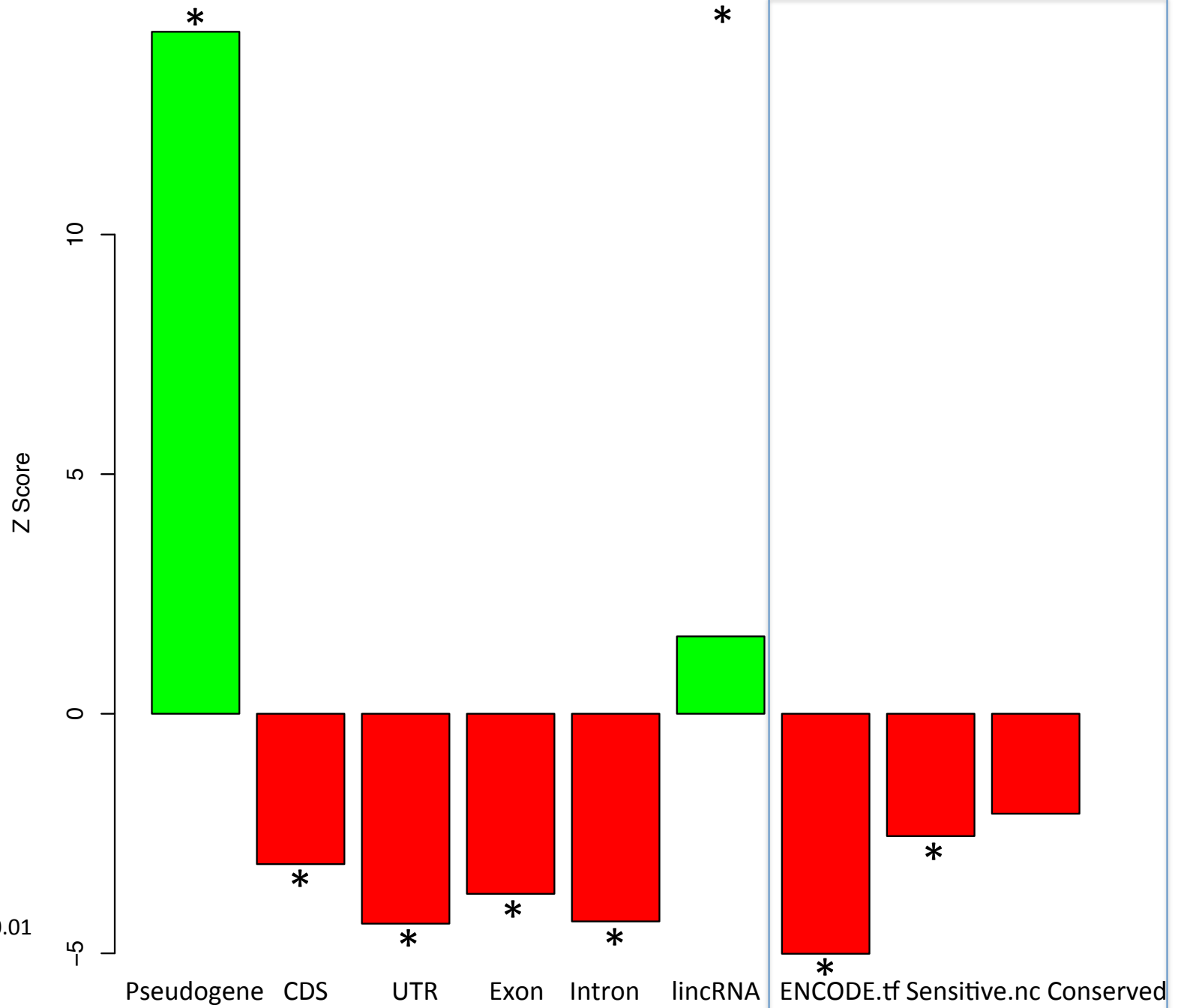
9/17/2014

# Different SV types
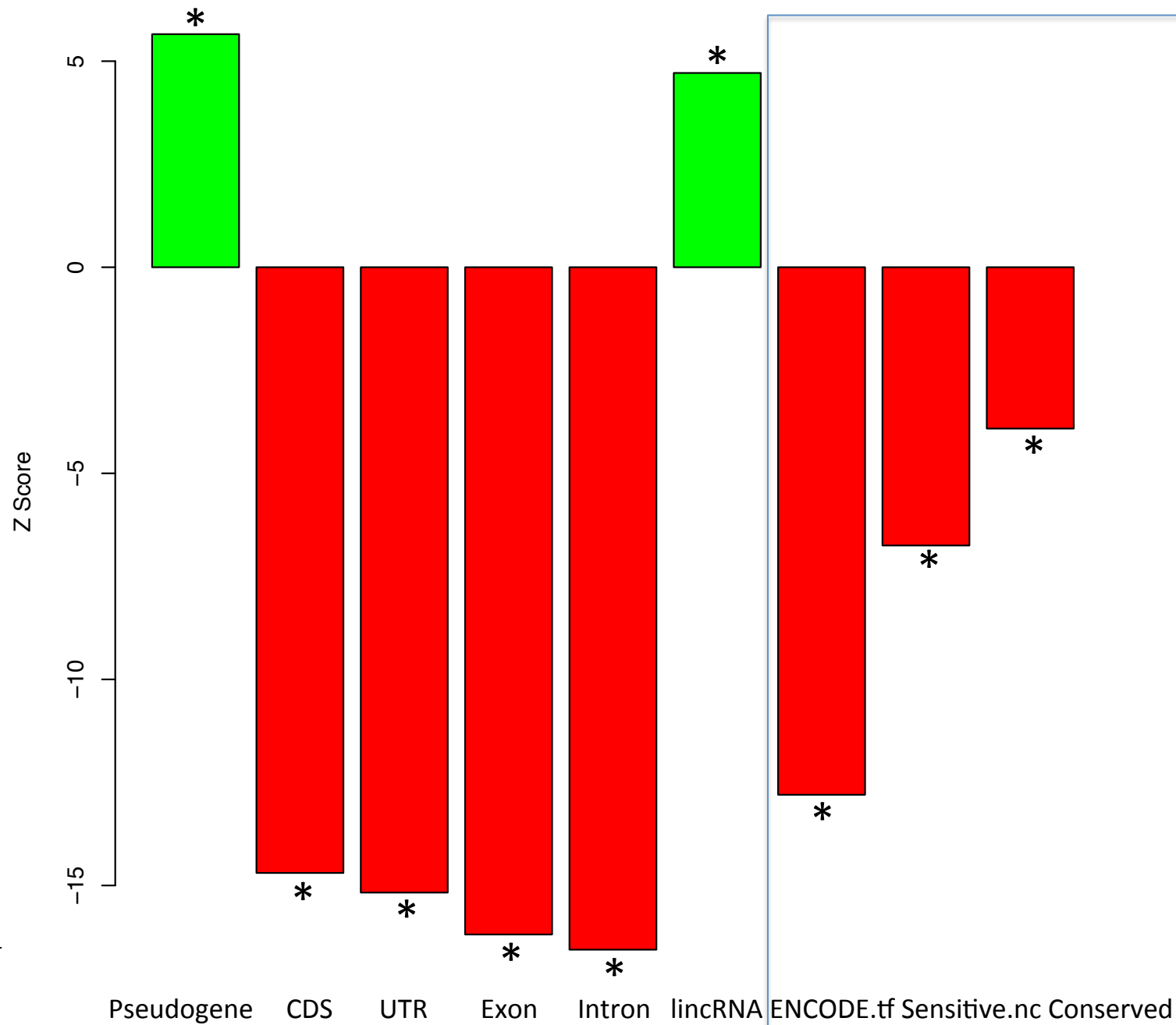
DEL

DUP

INV

CNV

INS:MT

INS:ME:SVA

INS:ME:LINE1
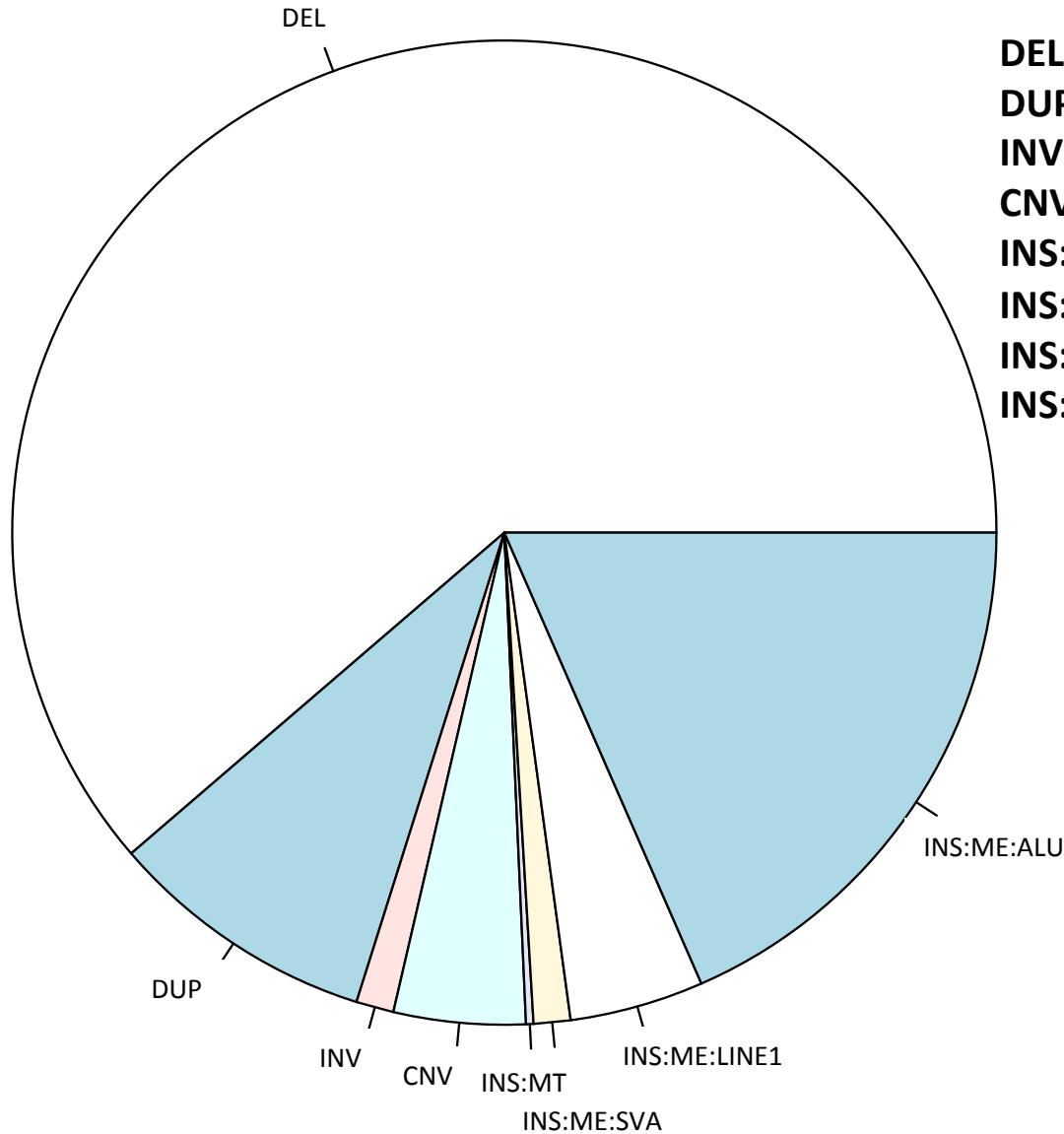
INS:ME:ALU

**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
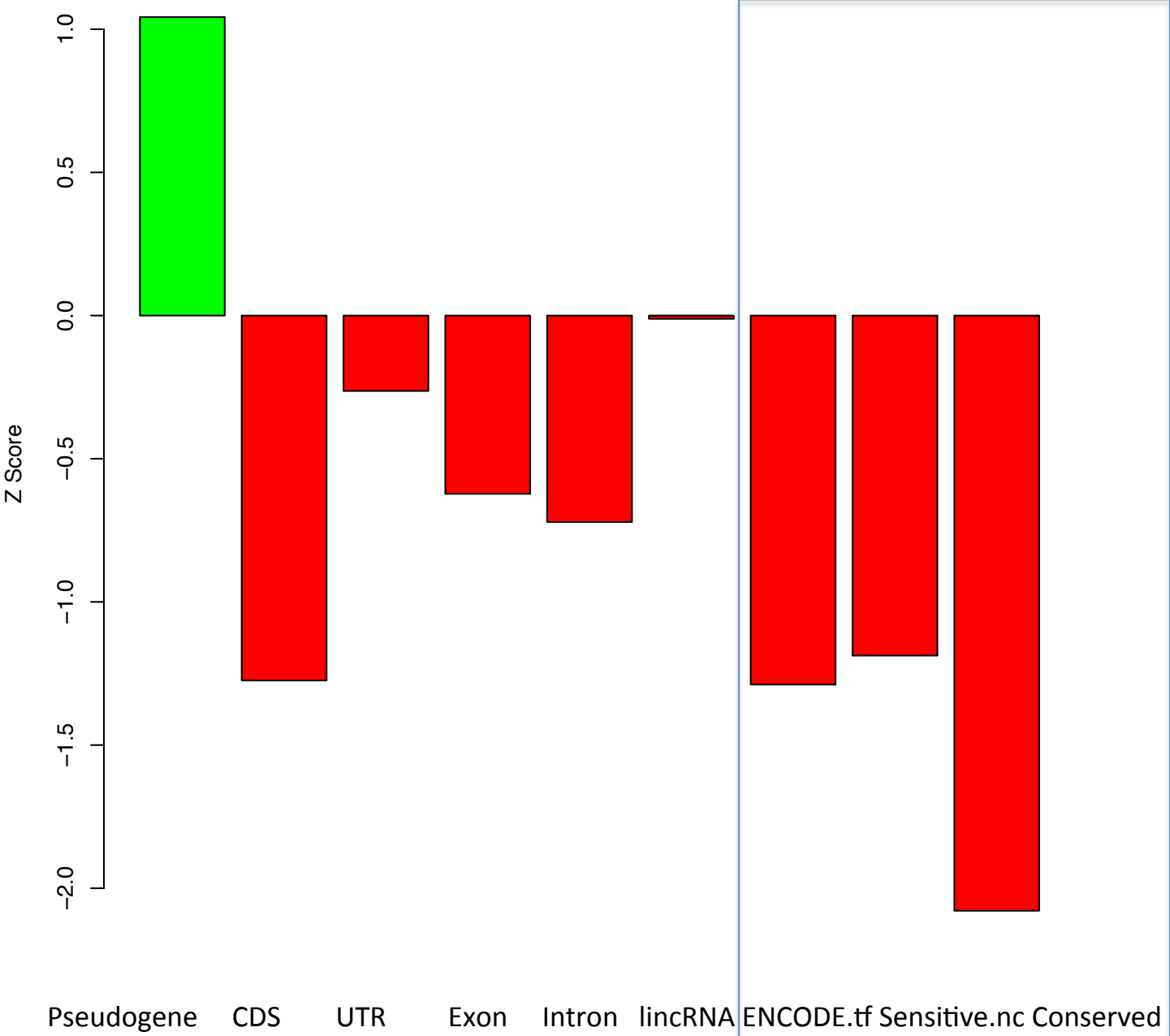
CNV overlap with genomic elements

**DEL overlap with genomic elements**

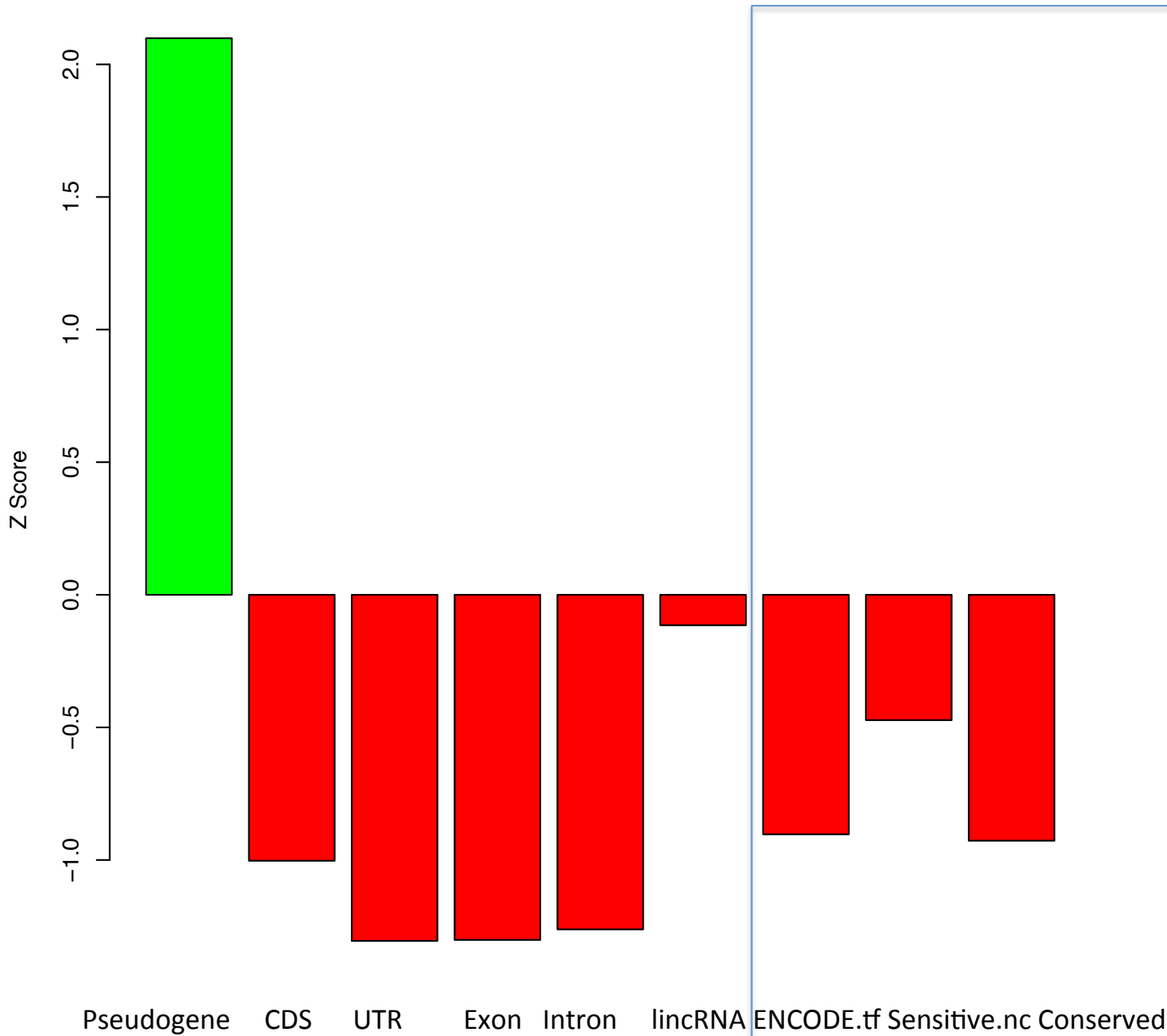Z Score

5

0

−5

−10

−15

* pvalue < 0.01

Pseudogene   CDS   UTR   Exon   Intron   lincRNA   ENCODE.tf   Sensitive.nc   Conserved

**DUP overlap with genomic elements**

Z Score

* pvalue < 0.01

Pseudogene   CDS   UTR   Exon   Intron   lincRNA   ENCODE.tf   Sensitive.nc   Conserved
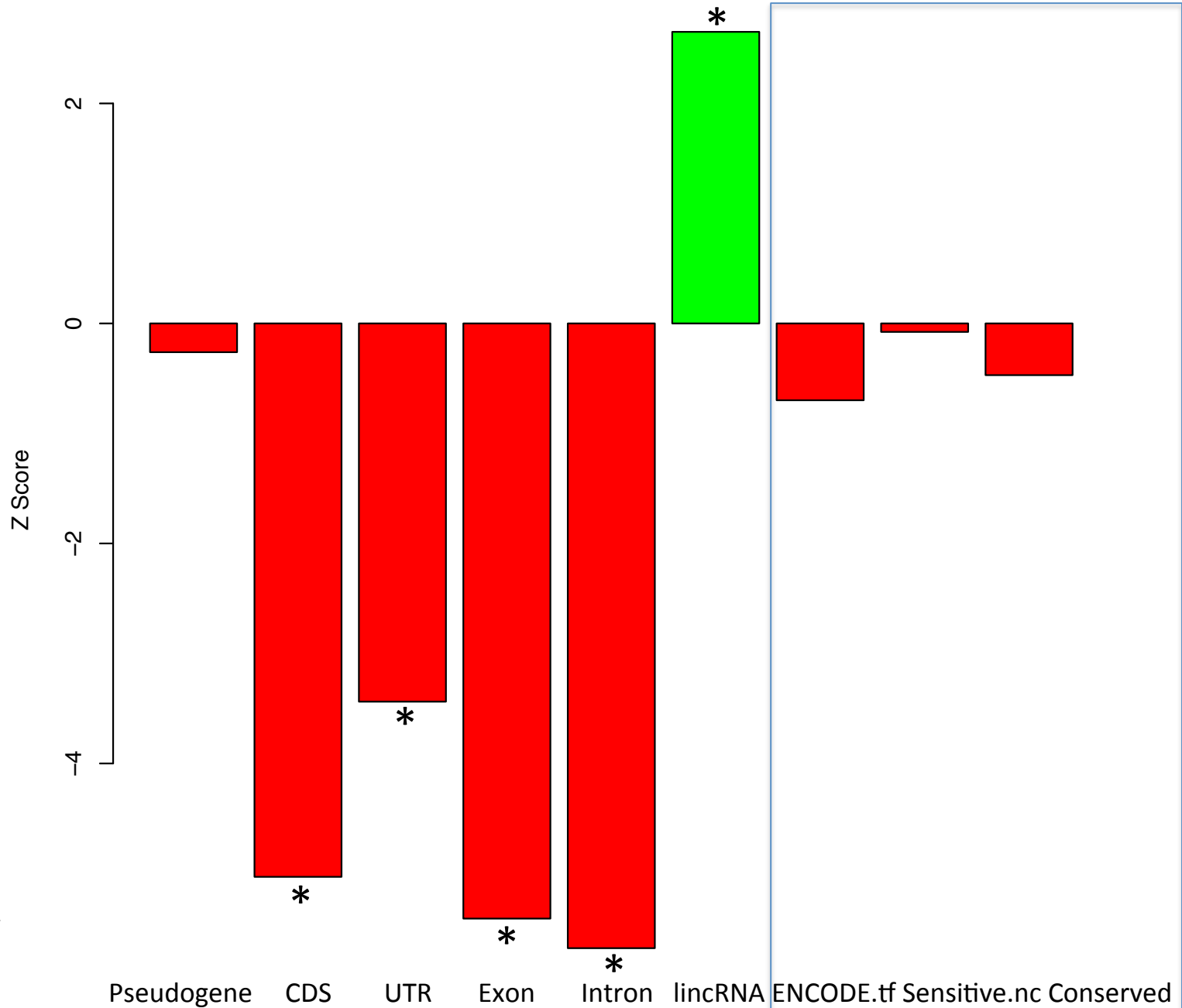
**INV overlap with genomic elements**

* pvalue < 0.01

**INS_ALU insertion position overlap with genomic elements**

Z Score

* pvalue < 0.01

Pseudogene  CDS  UTR  Exon  Intron  lincRNA  ENCODE.tf  Sensitive.nc  Conserved

**INS_LINE1 insertion position overlap with genomic elements**

Z Score

* pvalue < 0.01

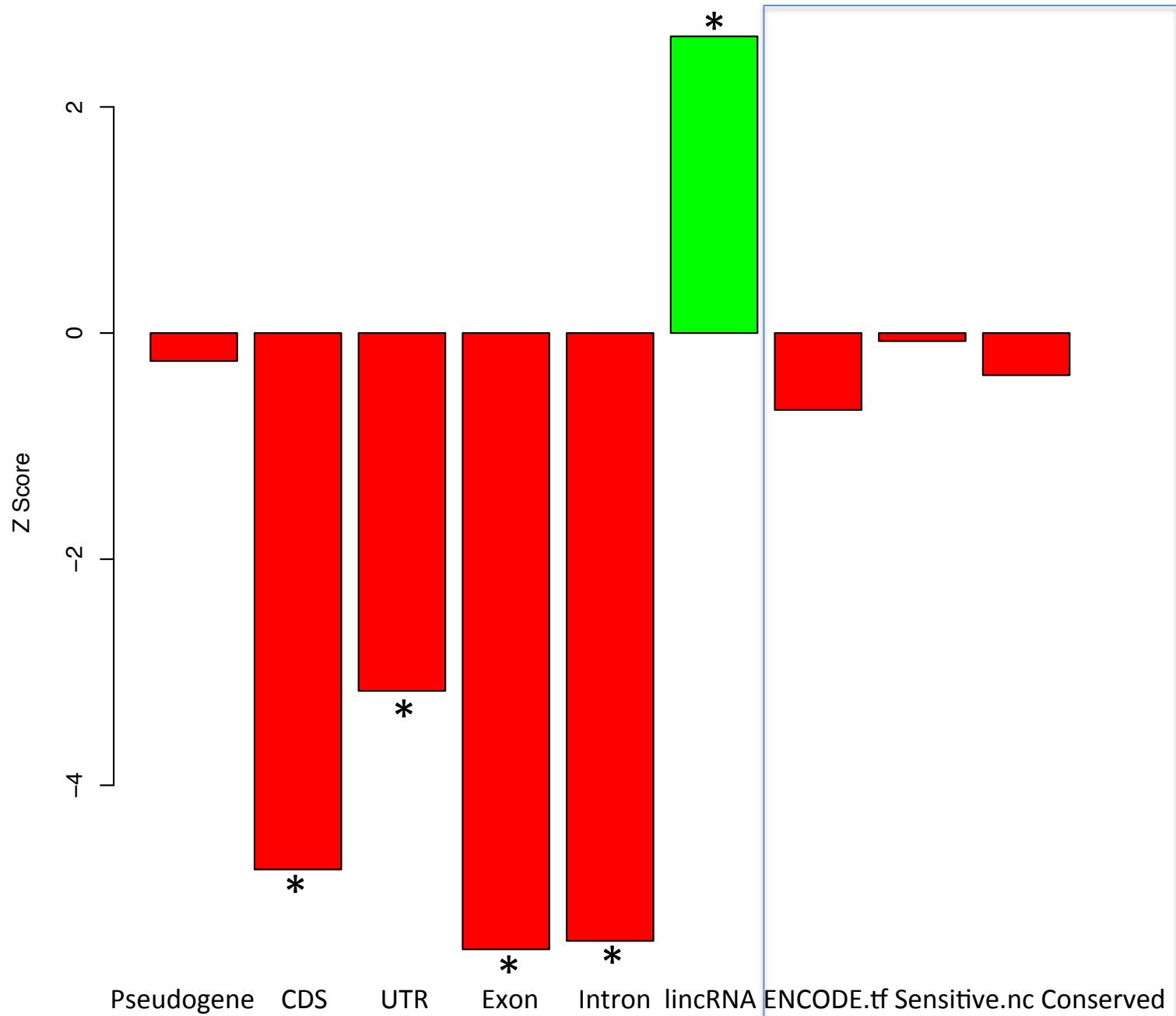Pseudogene  CDS  UTR  Exon  Intron  lincRNA  ENCODE.tf  Sensitive.nc  Conserved

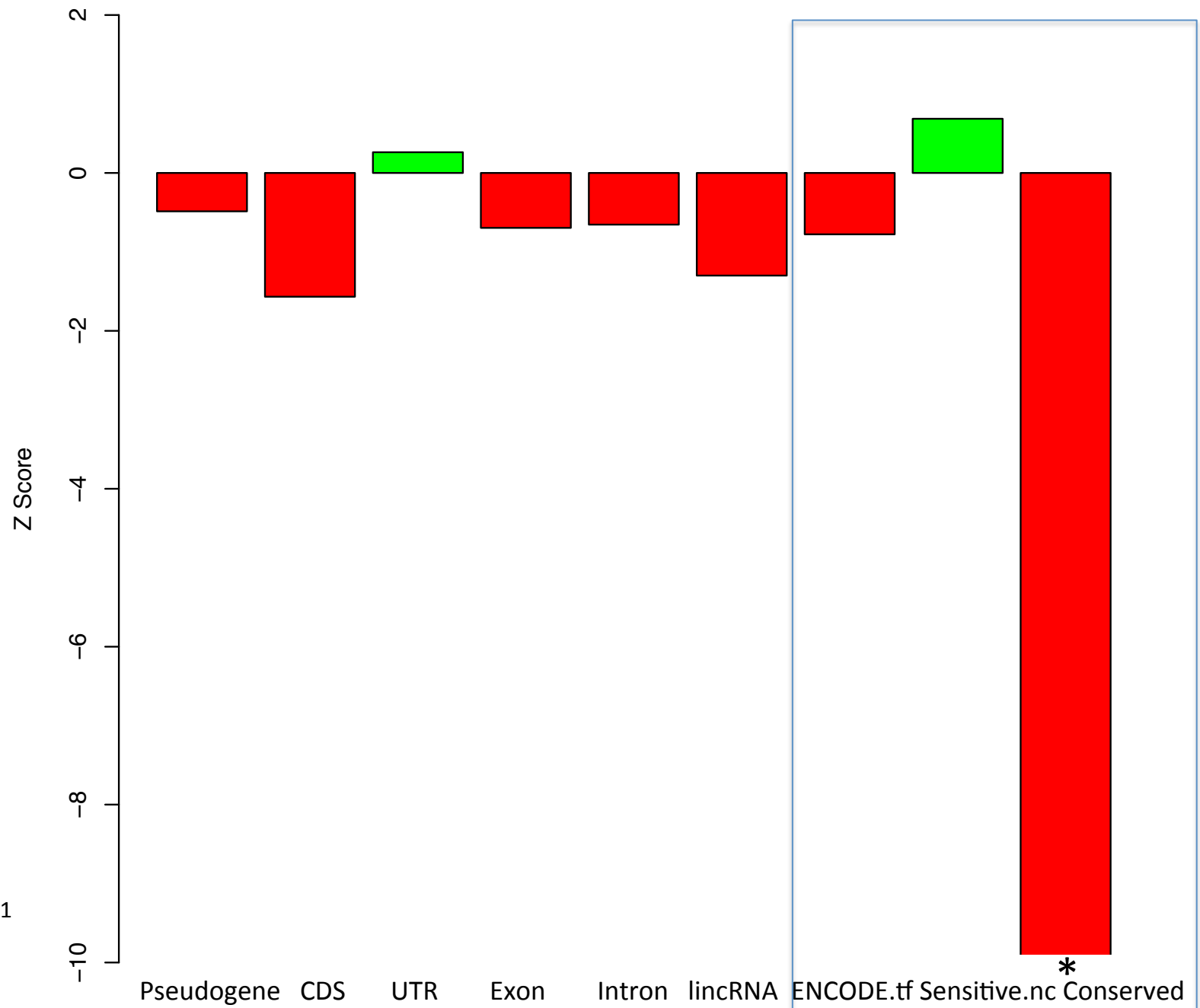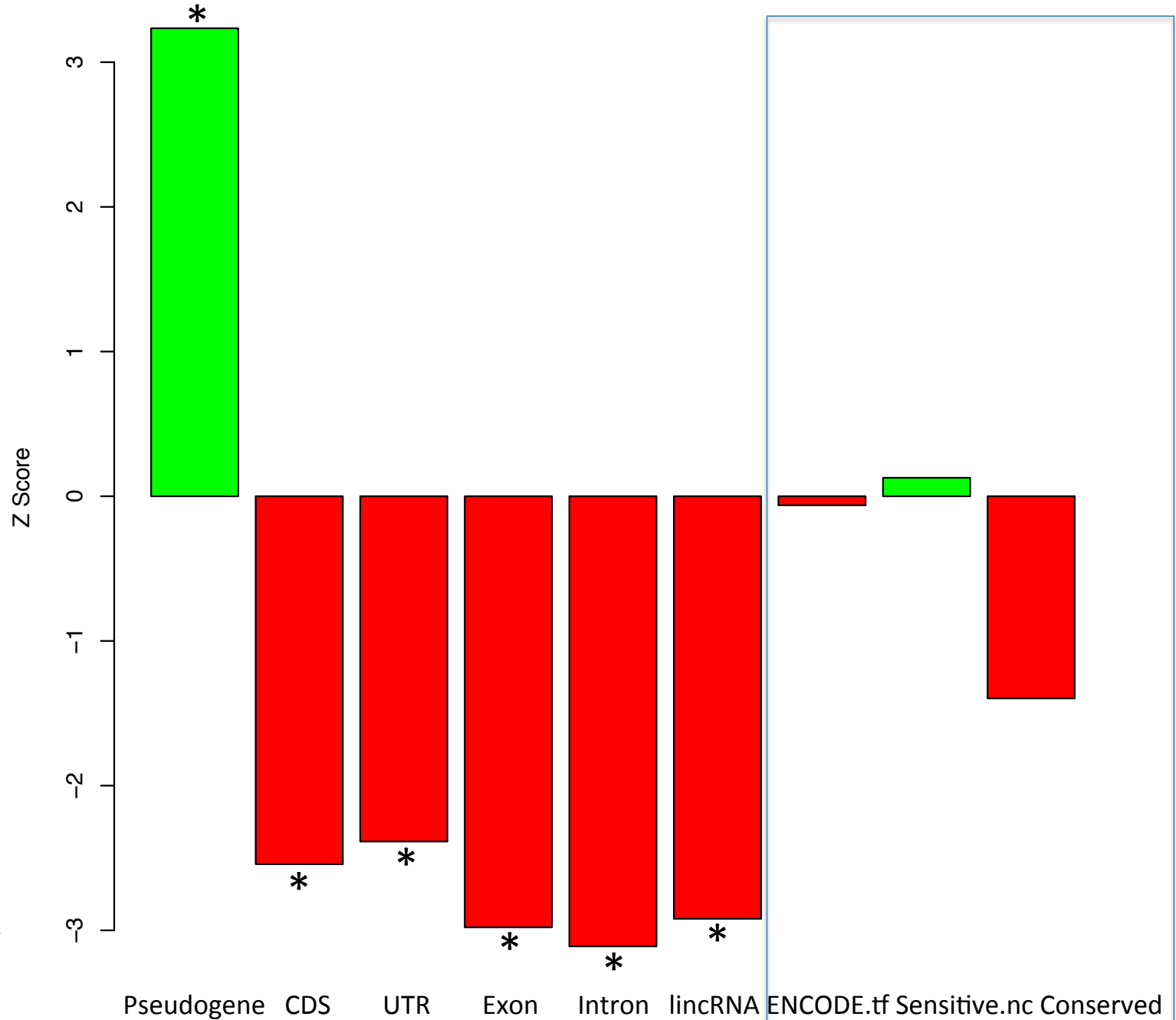**INS_MT insertion position overlap with genomic elements**

* pvalue < 0.01

**INS_SVA insertion position overlap with genomic elements**

Z Score

* pvalue < 0.01

Pseudogene  CDS  UTR  Exon  Intron  lincRNA  ENCODE.tf  Sensitive.nc  Conserved