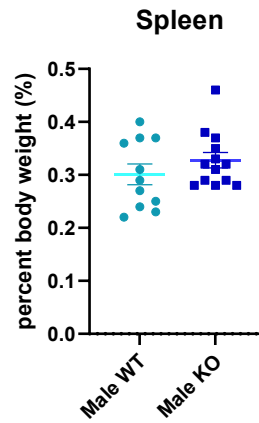
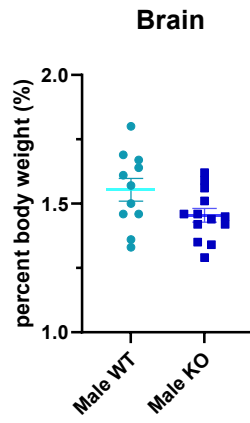
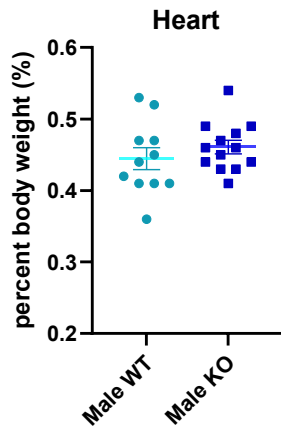
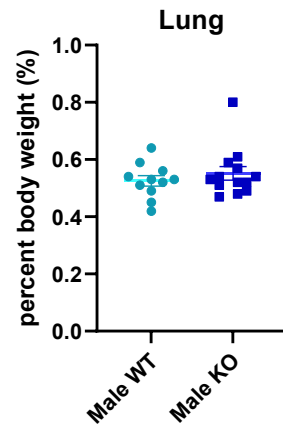
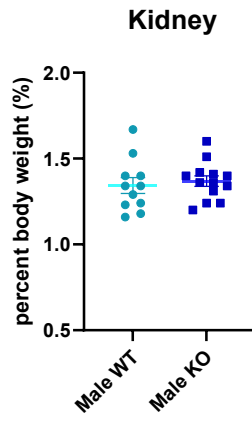
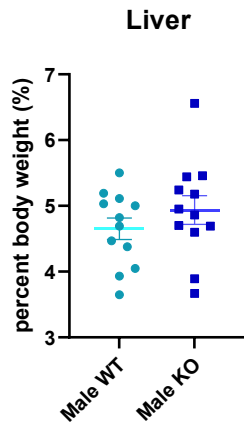
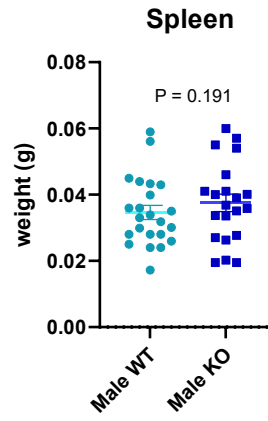
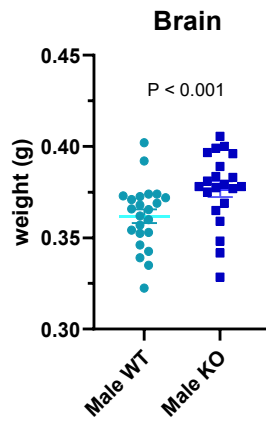
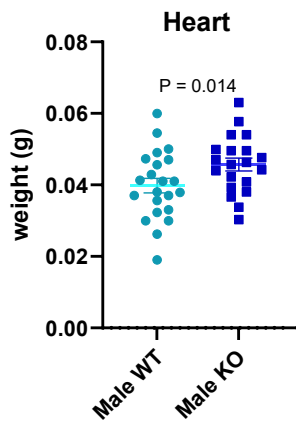
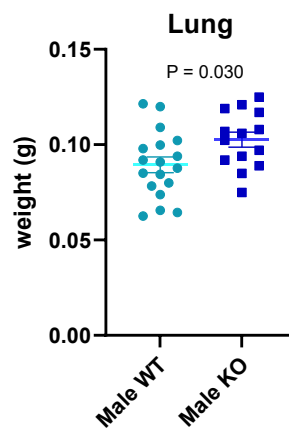
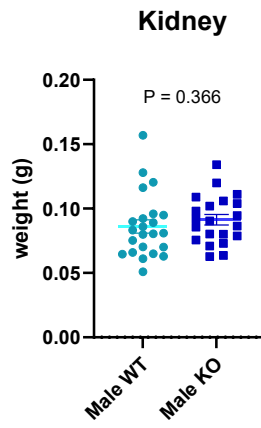
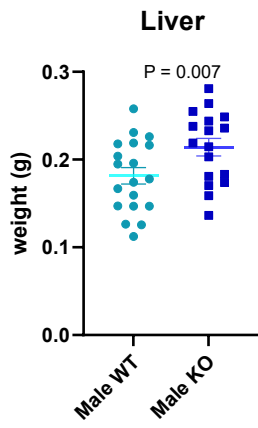
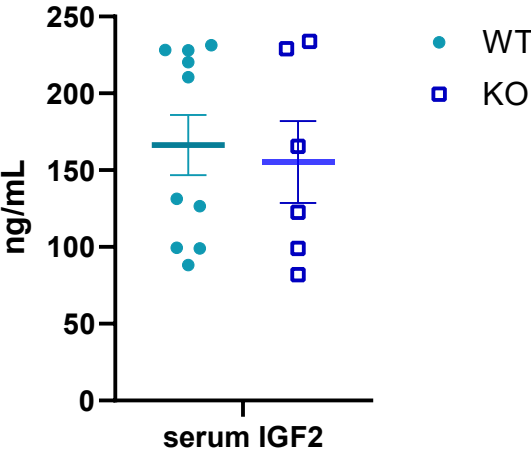


| Age | Reference Range (ng/mL) | Reference Mean (ng/mL) |
|-------------|-------------------------|------------------------|
| Prepubertal | 258-882 | 570 |
| Pubertal | 273-892 | 583 |
| Adults | 333-967 | 650 |

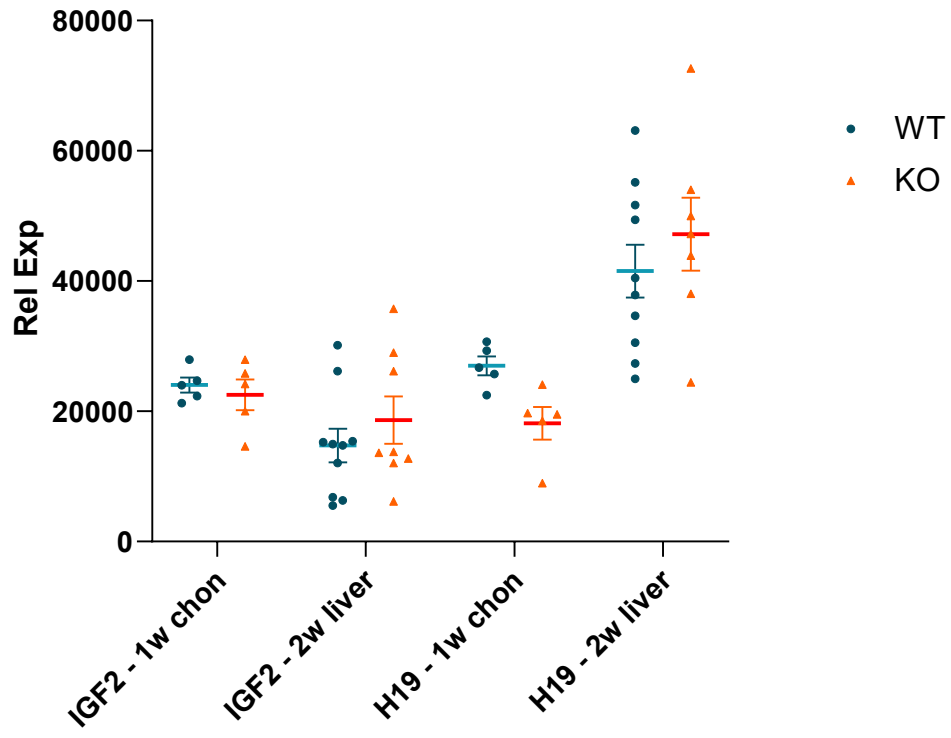




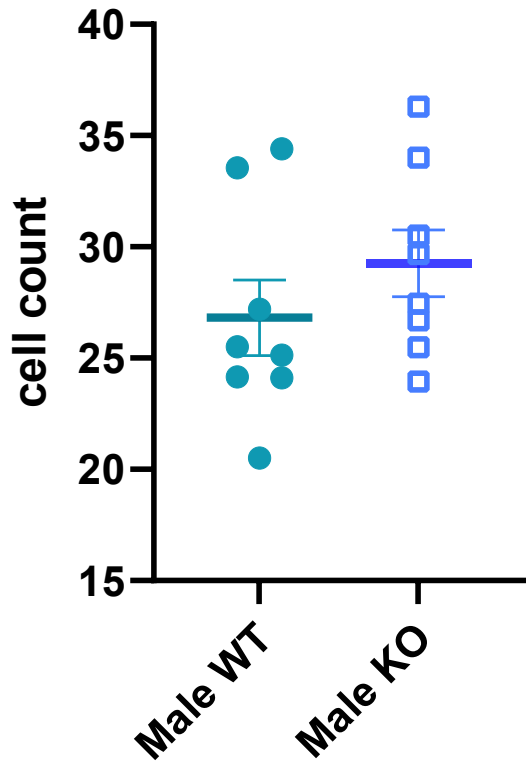
2wk old mice



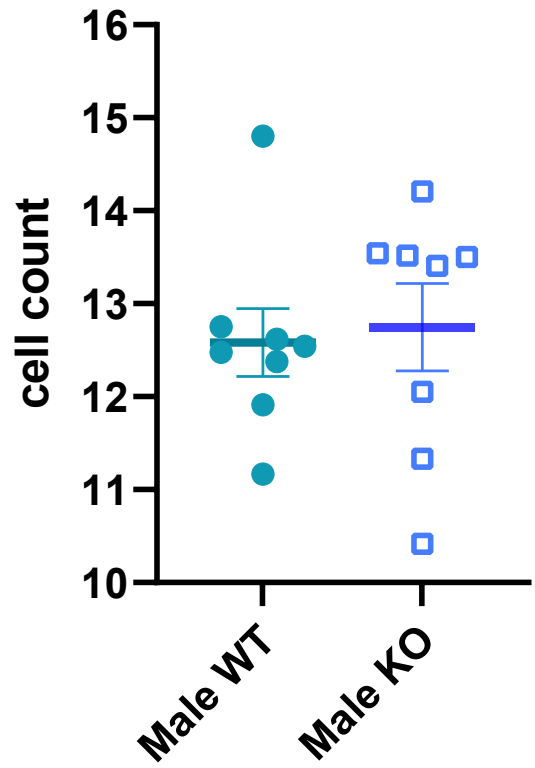
IGF2/H19 expression



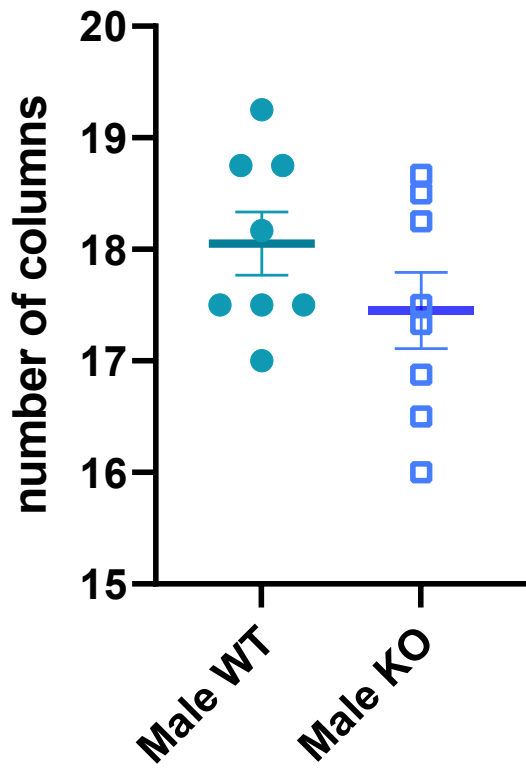
PZ cells/column



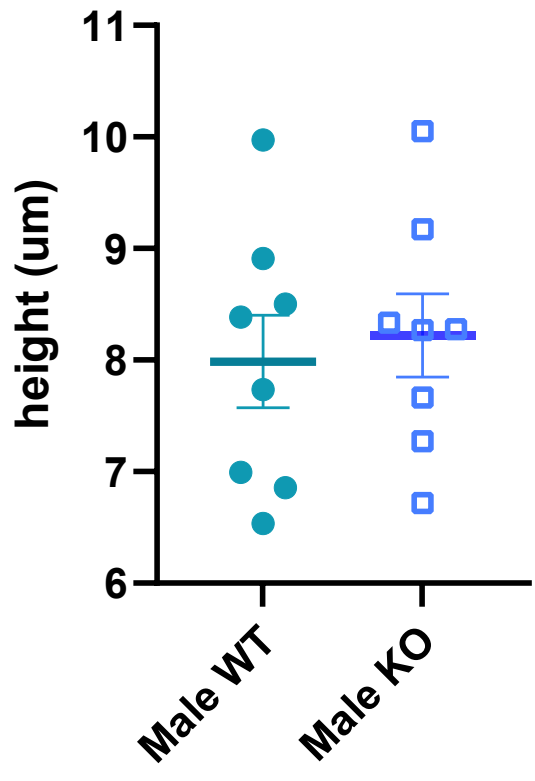
HZ cells/column



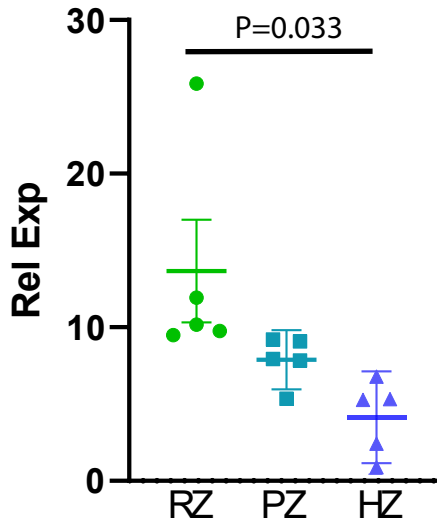
chon col/0.5mm



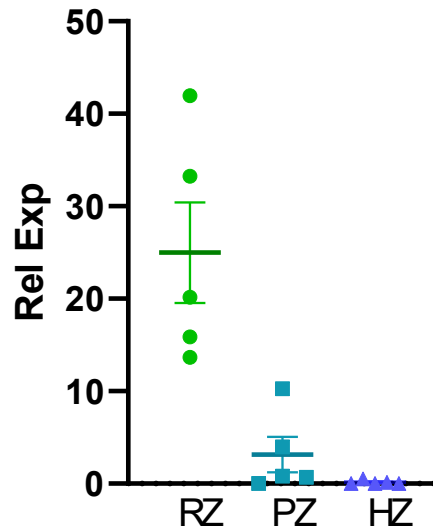
PZ cell height



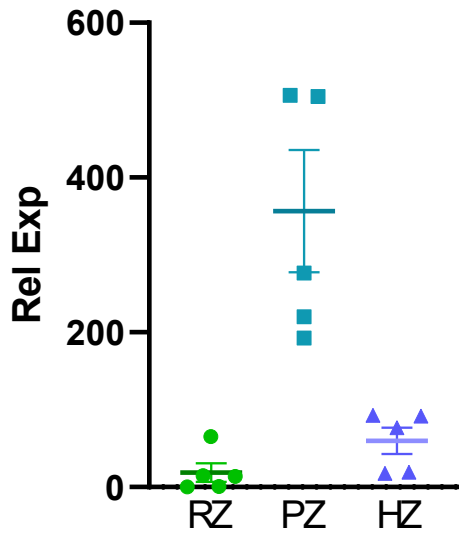
Spin4



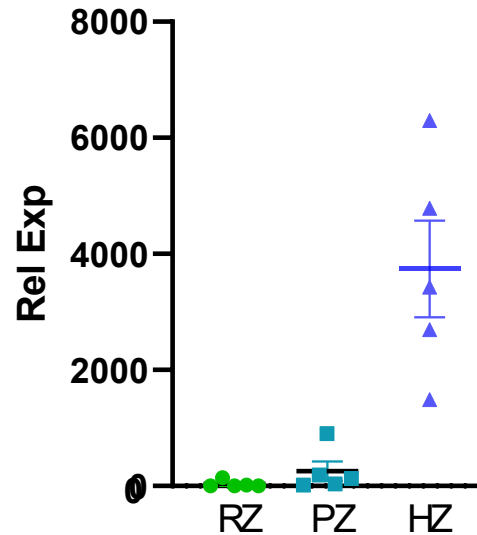
Pthlh (RZ marker)



Gdf10 (PZ marker)



Col10a1 (HZ marker)



Human subject

unaffected

```
tudor domain 1 CCT GAG AGA GTG CCA ACT CCT CGT ATC GAT TCA CGA CTG GCA GAT TCC CTG ATT GGC ... tudor domain 2
Pro Glu Arg Val Pro Thr Pro Arg Ile Asp Ser Arg Leu Ala Asp Ser Leu Ile Gly ...
```

proband

missing AG

```
tudor domain 1 CCT GAG AGT GCC AAC TCC TCG TAT CGA TTC ACG ACT GGC AGA TTC CCT GAT TGG CAA GGC AGT GGA GCA TGT GTT TGA
Pro Glu Ser Ala Asn Ser Ser Tyr Arg Phe Thr Thr Gly Arg Phe Pro Asp Trp Gln Gly Ser Gly Ala Cys Val STOP
```

Mouse models

Mouse Spin4 (amino acid 1-45)

```
ATG TCT CCT CCG ACT GTG CCT CCT ACT GGG GTG GAT GGT GTG TCC GCG TAC CTG ATG AAG AAA AGG CAC ACT CAC AAG AAG CAG
Met Ser Pro Pro Thr Val Pro Pro Thr Gly Val Asp Gly Val Ser Ala Tyr Leu Met Lys Lys Arg His Thr His Lys Lys Gln
CGC CGT AAG CCC ACT TTC CTC ACT CAT AGG AAC ATC GTG GGC TGC CGC ATT ... tudor domain 1
Arg Arg Lys Pro Thr Phe Leu Thr His Arg Asn Ile Val Gly Cys Arg Ile ...
```

Mouse Spin4 (long-deletion): missing all 3 Tudor domains

```
ATG TCT CCT CCG ACT GTG CCT CCT ACT GGG GTG GAT GGT GTG TCC GCG TAC AGC TGT CGA GCA CCT CCC CAG GCT CCT GTT CAG
Met Ser Pro Pro Thr Val Pro Pro Thr Gly Val Asp Gly Val Ser Ala Tyr Ser Cys Arg Ala Pro Pro Gln Ala Pro Val Gln
CTG TGG GCA AGC AGG TGG AAC ACG CCA AAG ACG ACG GGT CCA AGA GAA CTG GGA TCT TTA TTC ATC AGG TGG TAG
Leu Trp Ala Ser Arg Trp Asn Thr Pro Lys Thr Thr Gly Pro Arg Glu Leu Gly Ser Leu Phe Ile Arg Trp STOP
```

Mouse Spin4 (amino acid 165-215)

```
TAC ACC CTG CTG GAC GAC TAC AAA GAT GGT GAC CTG CGT ATC ATT CCA GAT TCC AAC TAC TAC TTC CCT GCA GCT GAA CAG GAG
Tyr Thr Leu Leu Asp Asp Tyr Lys Asp Gly Asp Leu Arg Ile Ile Pro Asp Ser Asn Tyr Tyr Phe Pro Ala Ala Glu Gln Glu
CCT GGG GAG GTG CTC GAC AGC CTT GTG GGC AAG CAG GTG GAA CAC GCC AAA GAC GAC GGG TCC AAG AGA ... tudor domain 3
Pro Gly Glu Val Leu Asp Ser Leu Val Gly Lys Gln Val Glu His Ala Lys Asp Asp Gly Ser Lys Arg ...
```

Mouse Spin4 (short-deletion): missing Tudor domain 3

```
TAC ACC CTG CTG GAC GAC TAC AAA GAT GGT GAC CTG CGT ATC ATT CCA GAT TCC AAC TAC TAC TTC CCT GCC AAG CCA TCT GTC
Tyr Thr Leu Leu Asp Asp Tyr Lys Asp Gly Asp Leu Arg Ile Ile Pro Asp Ser Asn Tyr Tyr Phe Pro Ala Lys Pro Ser Val
TAC TTC ATT AAG TTT GAT GAT GAT ATT CAC ATT TAT GTC TAT GGC TTG GTG AAA ACC CCG TAA
Tyr Phe Ile Lys Phe Asp Asp Asp Ile His Ile Tyr Val Tyr Gly Leu Val Lys Thr Pro STOP
```