

**Regulatory features of the spinocerebellar ataxia
type 2 gene (*ATXN2*).**

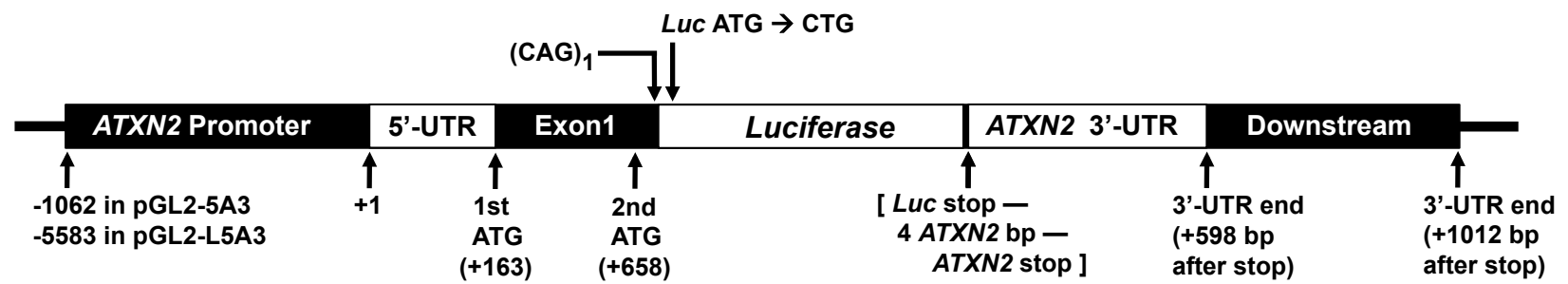


FIG 1

A

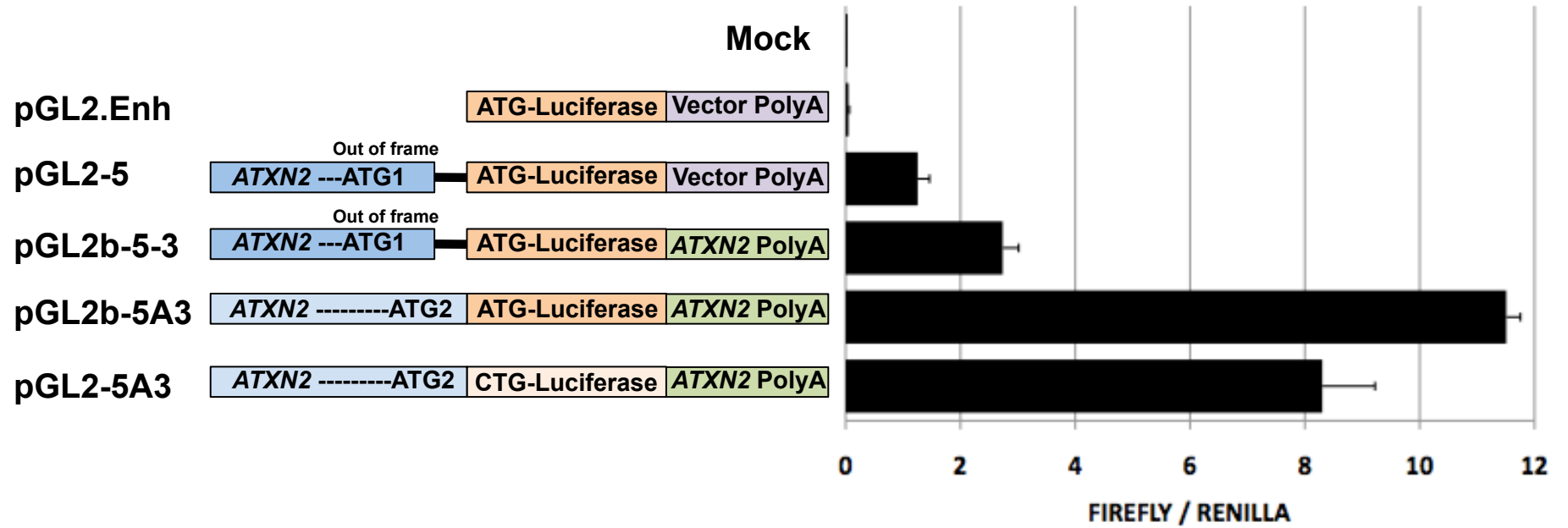
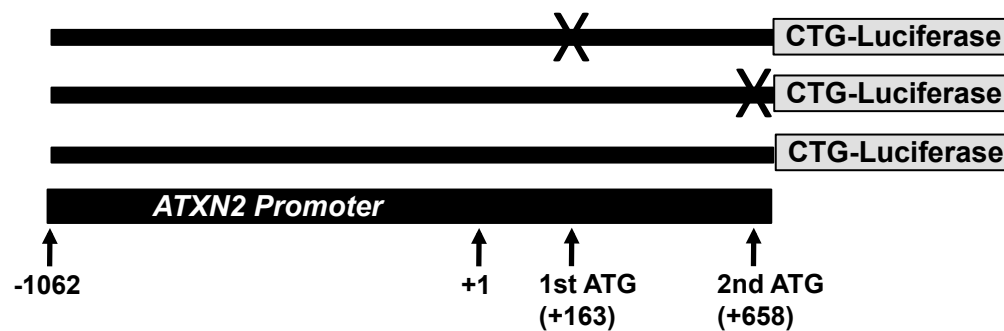
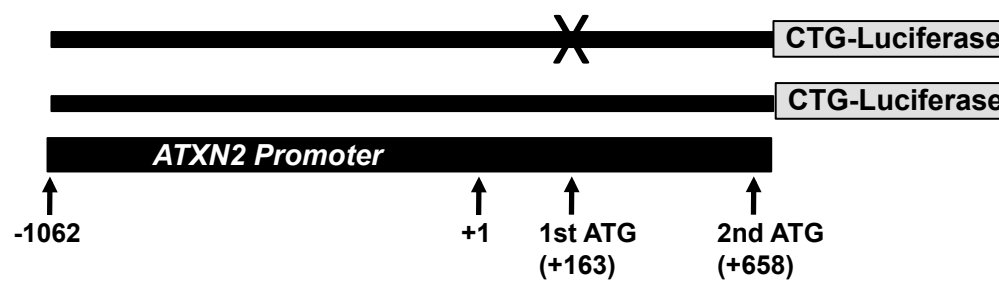


Fig 2A

B



ADD CHART

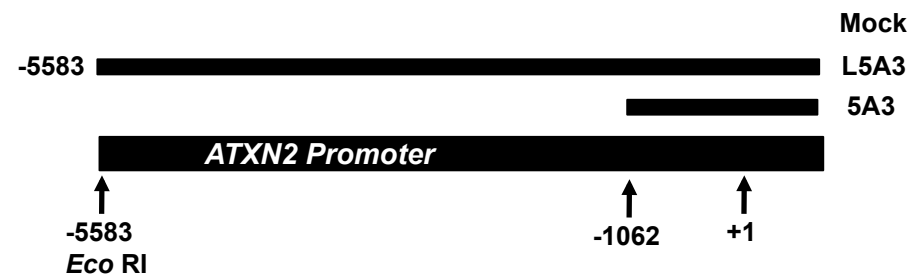


ADD CHART

Fig 2B

Western blot with ATGs mutated

A



ADD CHART

Fig 3A

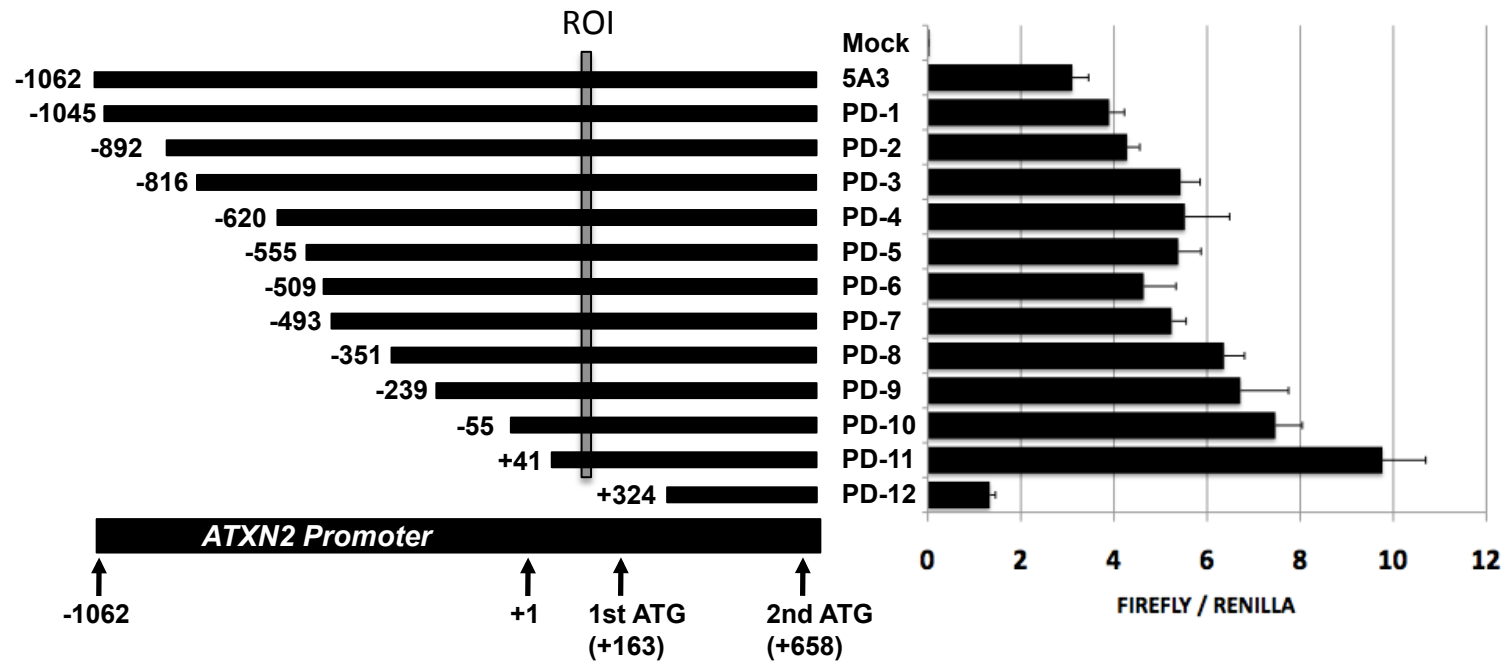
B

Fig 3B

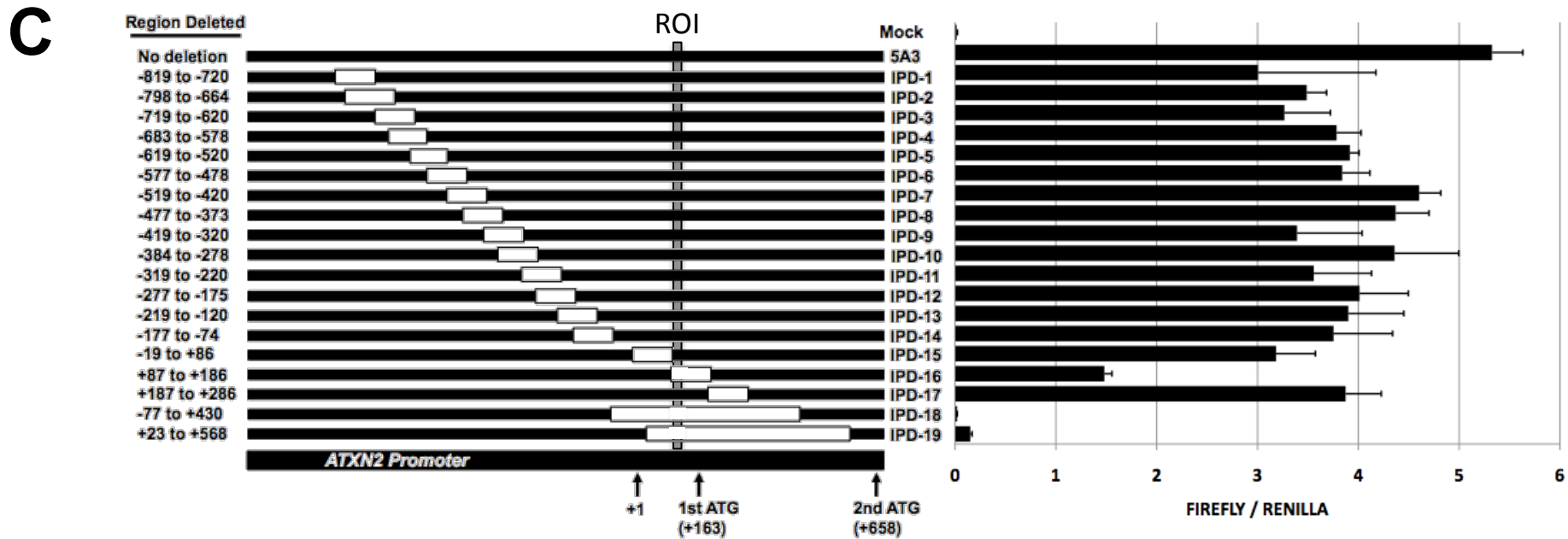
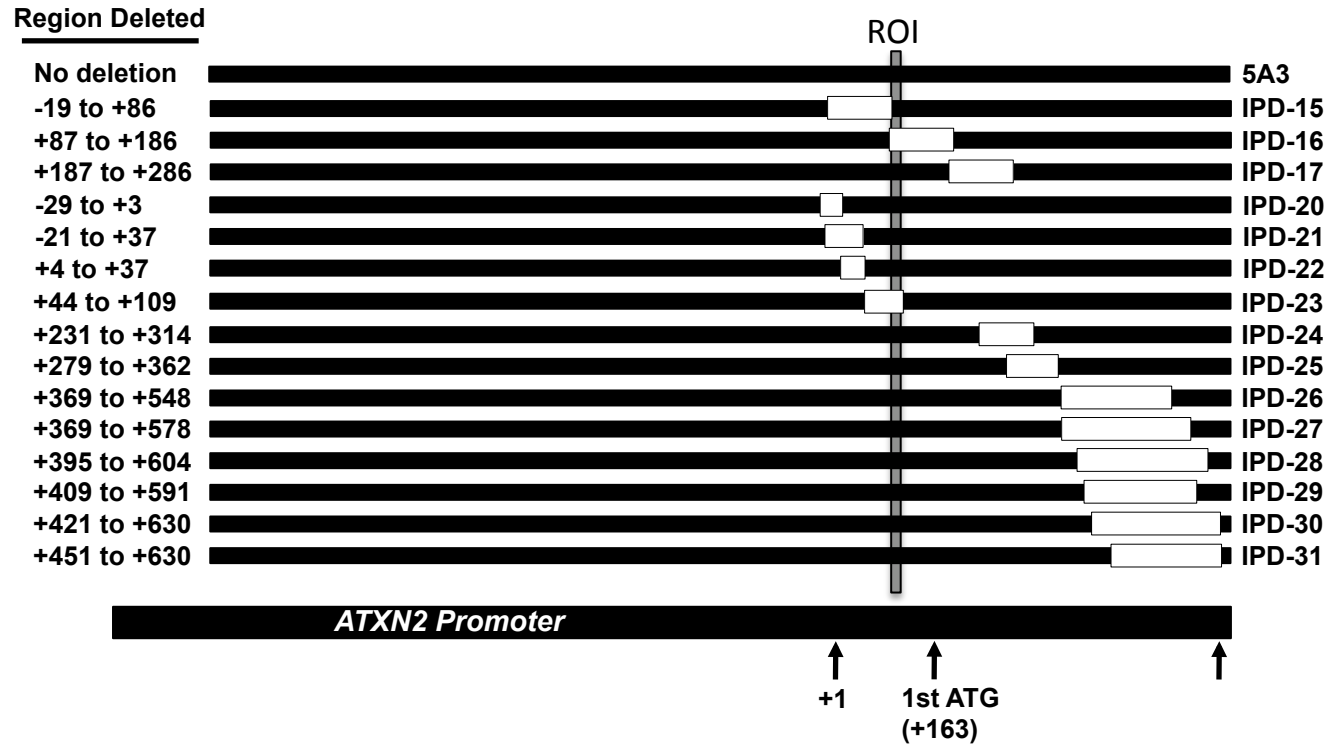


Fig 3C

D



ADD Chart

Fig 3D

A

DICE (-) acgttctgcttccgtctgacccctccgacttccggtaaagagtccctatccgcacctccgctcccaccgggc
 ETSF (-) acgttctgcttccgtctgacccctccgacttccggtaaagagtccctatccgcacctccgctcccaccgggc
 FKHD (+) acgttctgcttccgtctgacccctccgacttccggtaaagagtccctatccgcacctccgctcccaccgggc
 HOMF (+) acgttctgcttccgtctgacccctccgacttccggtaaagagtccctatccgcacctccgctcccaccgggc
 STAT (+) acgttctgcttccgtctgacccctccgacttccggtaaagagtccctatccgcacctccgctcccaccgggc
 STAT (-) acgttctgcttccgtctgacccctccgacttccggtaaagagtccctatccgcacctccgctcccaccgggc
 "SO7" acgttctgcttccgtctgacccctccgacttccggtaaagagtccctatccgcacctccgctcccaccgggc
 "WT" acgttctgcttccgtctgacccctccgacttccggtaaagagtccctatccgcacctccgctcccaccgggc

Primer MutR1 5'-ccgtctgacccctccgactcgcaggtaaagagtccc-3' → ETSF & FKHD remain
 Primer MutR2 5'-ccgtctgacccctc-----gagtcctatccgc-3' → All eliminated

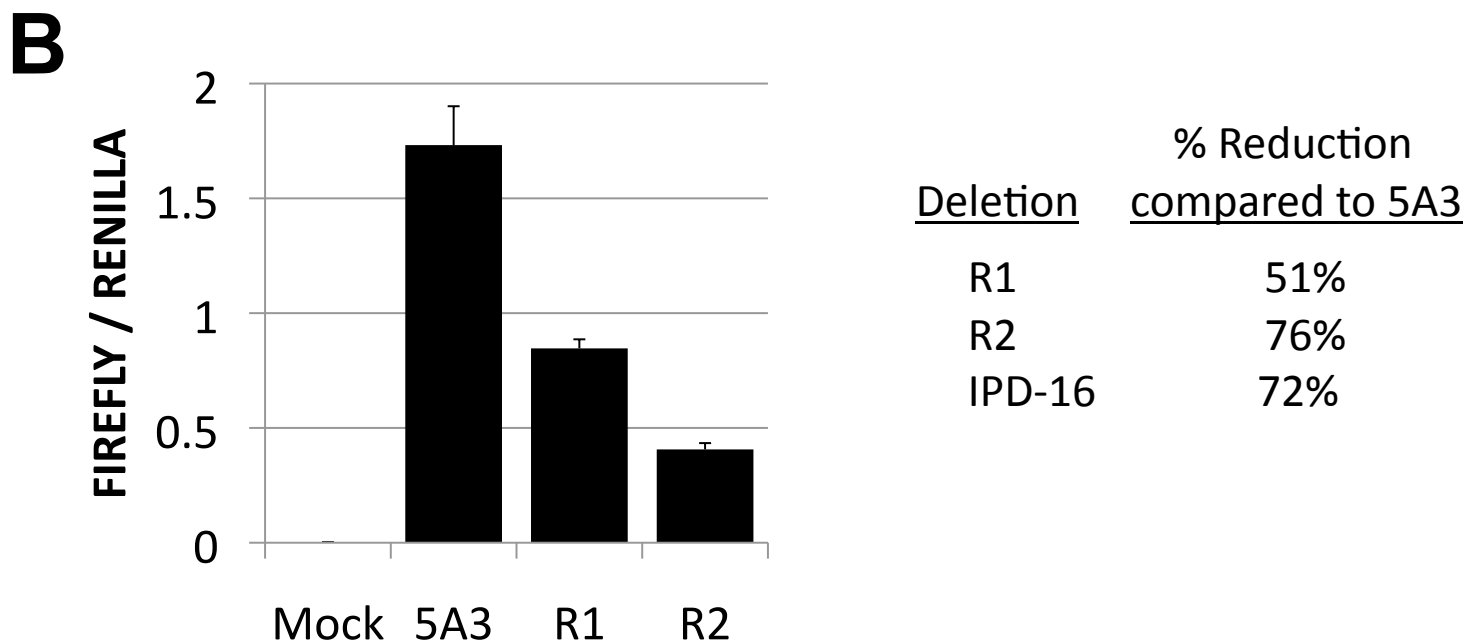
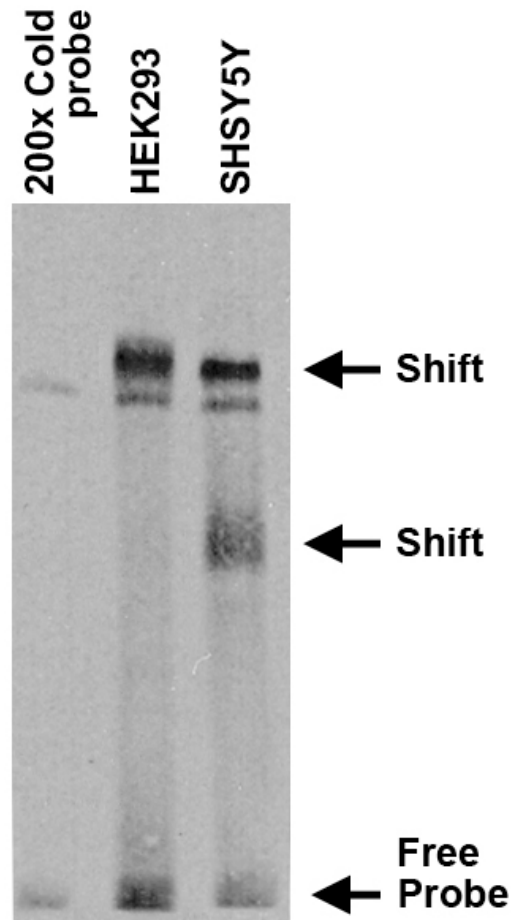


Fig 4

EMSA data



Probe = "WT"

EMSA with Supershift seen for NERFc antibody

9/8/2010

10 min cold probe incubation
Then added hot probe and
Incubated additional 60 min

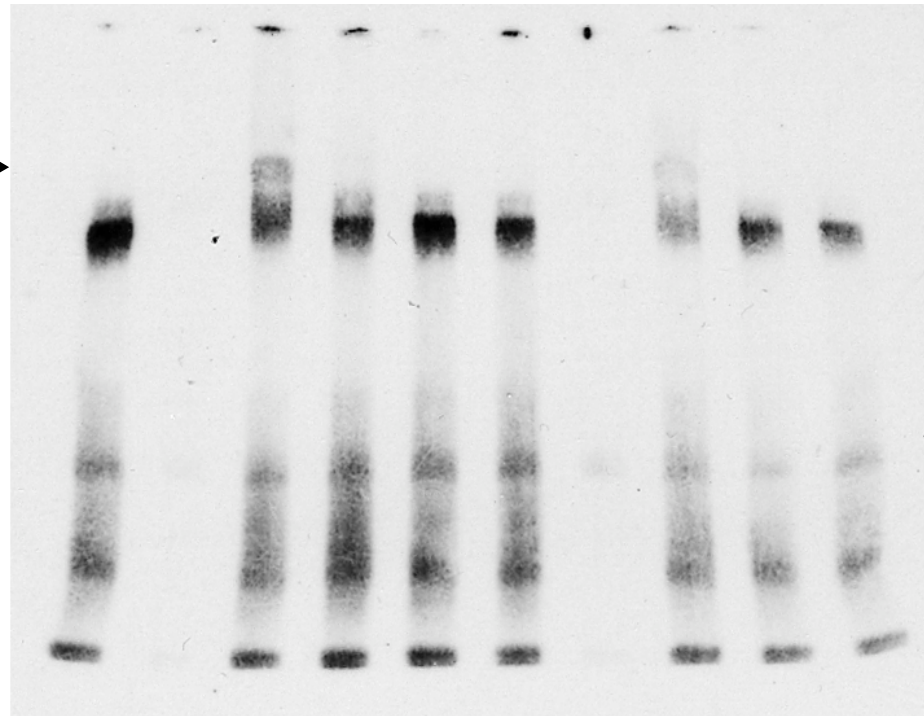
10 min cold probe incubation
Then added hot probe and
Incubated additional 20 min (my usual method)

No Antibody	200x Cold Probe	NERFc Antibody	NERFv Antibody	STAT3 Antibody	No Antibody	200x Cold Probe	NERFc Antibody	NERFv Antibody	STAT3 Antibody
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The reactions between
Left and right are
identical but they have
2.5 times the amount of
hot probe and 50%
more nuclear lysate

Supershift for ELF2 →

6% gel run for
3 hours at 175 V



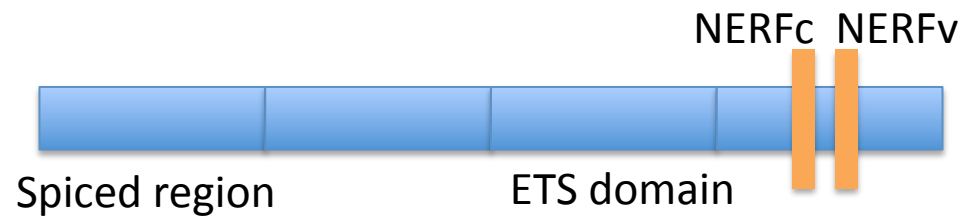
ELF2

E74-like transcription factor isoform 2

NERF = New ETS related factor

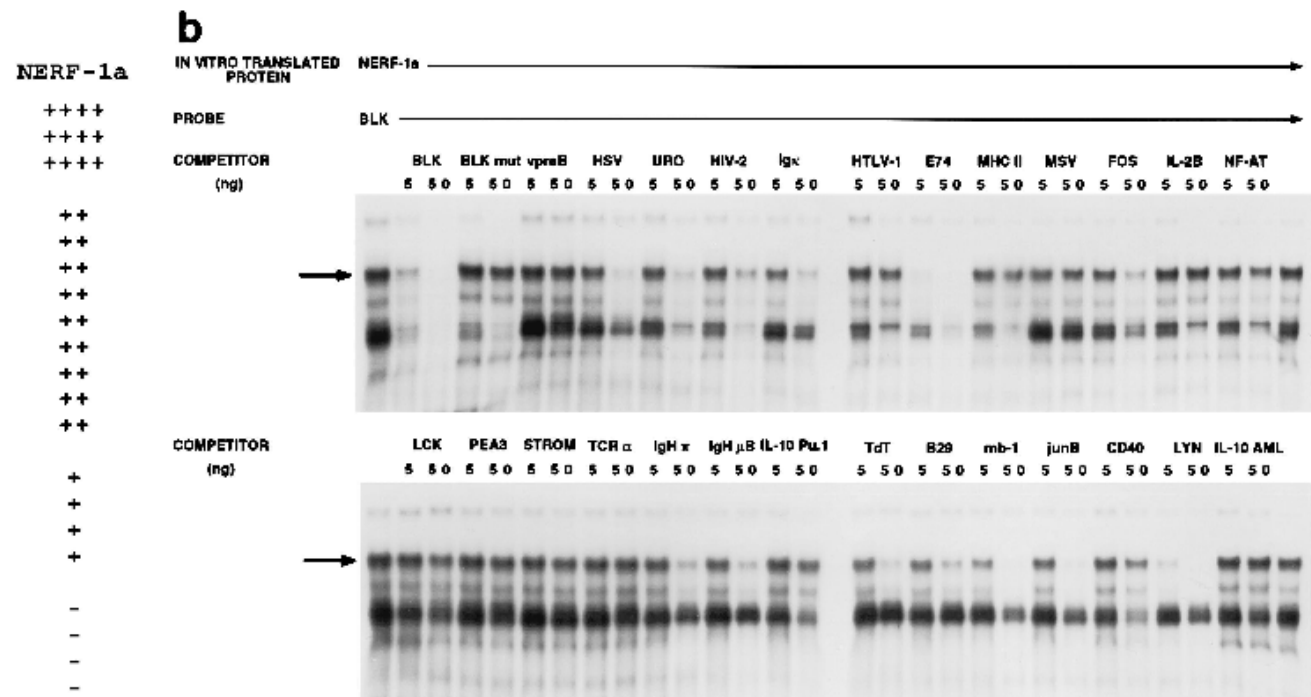
NERF= NERF1a NERF1b NERF2
splicing in the N term

Member of the ETS family



From Oettgen et al., 1996, characterization of NERF...

<i>MutR1</i>	TACCTCGAGTCG
<i>ATXN2</i>	TACCGGAAGTCG
a	
ENHANCER	BINDING SITE
blk	TCCAGGAAGTAT
lyn	ACCAGGAAGTAG
E74	AACCGGAAGTAA
HSV ICP4	GAACGGAAGCGG
IgH Enh. π site	GGCAGGAAGCAG
IgH Enh. μ B site	TTGGGGAAGGGA
TdT	GGCAGGAAGCTG
mb-1	AACAGGAAGTGA
junB	GTCAGGAAGCGC
UROKINASE	AGGAGGAATGA
B29	GGCAGGAAGGGG
HIV-2 LTR CD3R	GACAGGAACAGC
Ig κ 3' enh.	TTGAGGAAC TGA
LCK Prom.	GGCAGGAAGCTT
c-FOS SRE	CACAGGATGTCC
IL-10 Pu.1	ATGCAGAAGTTC
IL-2 NF-AT	AGGAGGAAAAC
HTLV-1 LTR	GGGAGGAAATGG
STROMELYSIN Prom.	AGCAGGAAGCAT
MSV LTR	GACCGGAAGCGC
CD40	CCCAGGAAGAGG
MHC class II Prom.	GTGAGGAACCAA
Polyoma PEA3	AGCAGGAAGTGA
IL-2 IL-2B	AAGAGGAAAAT
TCR α Enh. T β 2	CAGAGGATGTGG
vpreB Prom.	GGGAGGAAGCAC
CONSENSUS	TCCAGGAAGTAN AA C



Expression based on information in the Human Protein Atlas www.proteinatlas.org/



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ELF2 expression profiles.

Gene data

Description: **E74-like factor 2 (ets domain transcription factor)**
Source acc: 3317 (HGNC Symbol)
Chromosome: 4:q31.1
Ensembl ID: **ENSG00000109381**

	Protein Ensembl ID	Transcript Ensembl ID	No of aa	Mw
Splice variant 1:	ENSP00000265495	ENST00000265495	581	63 kDa
Splice variant 2:	ENSP00000368868	ENST00000379550	593	64 kDa
Splice variant 3:	ENSP00000377782	ENST00000394235	581	63 kDa

alph. sort order

Normal Tissues - IHC

Adrenal gland	cortical cells	Lymph node	lymphoid cells outside reaction centra
Appendix	glandular cells		reaction center cells
	lymphoid tissue	Nasopharynx	respiratory epithelial cells
Bone marrow	bone marrow poietic cells	Oral mucosa	squamous epithelial cells
Breast	glandular cells	Ovary	follicle cells
Bronchus	respiratory epithelial cells		ovarian stromal cells
Cerebellum	cells in granular layer	Pancreas	exocrine glandular cells
	cells in molecular layer		islet cells
	purkinje cells	Parathyroid gland	glandular cells
Cerebral cortex	glial cells	Placenta	decidual cells
	neuronal cells		trophoblastic cells
Cervix, uterine	glandular cells	Prostate	glandular cells
	squamous epithelial cells	Rectum	glandular cells
Colon	glandular cells	Salivary gland	glandular cells
Corpus, uterine 1	cells in endometrial stroma	Seminal vesicle	glandular cells
	glandular cells	Skeletal muscle	myocytes
Corpus, uterine 2	cells in endometrial stroma	Skin	adnexal cells
	glandular cells		epidermal cells
Duodenum	glandular cells	Small intestine	glandular cells
Epididymis	glandular cells	Smooth muscle	smooth muscle cells
Esophagus	squamous epithelial cells	Soft tissue 1	mesenchymal cells
Fallopian tube	glandular cells	Soft tissue 2	mesenchymal cells
Gall bladder	glandular cells	Spleen	cells in red pulp
Heart muscle	myocytes		cells in white pulp
Hippocampus	glial cells	Stomach 1	glandular cells
	neuronal cells	Stomach 2	glandular cells
Kidney	cells in glomeruli	Testis	cells in seminiferous ducts
	cells in tubules		leydig cells
Lateral ventricle	glial cells	Thyroid gland	glandular cells
	neuronal cells	Tonsil	lymphoid cells outside reaction centra
Liver	bile duct cells		reaction center cells
	hepatocytes		squamous epithelial cells
Lung	alveolar cells	Urinary bladder	urothelial cells
	macrophages	Vagina	squamous epithelial cells
		Vulva/anal skin	squamous epithelial cells

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HPA006057

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Expression profiles - IF

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Validation Summary

IH IF PA WB

Search

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Protein expression

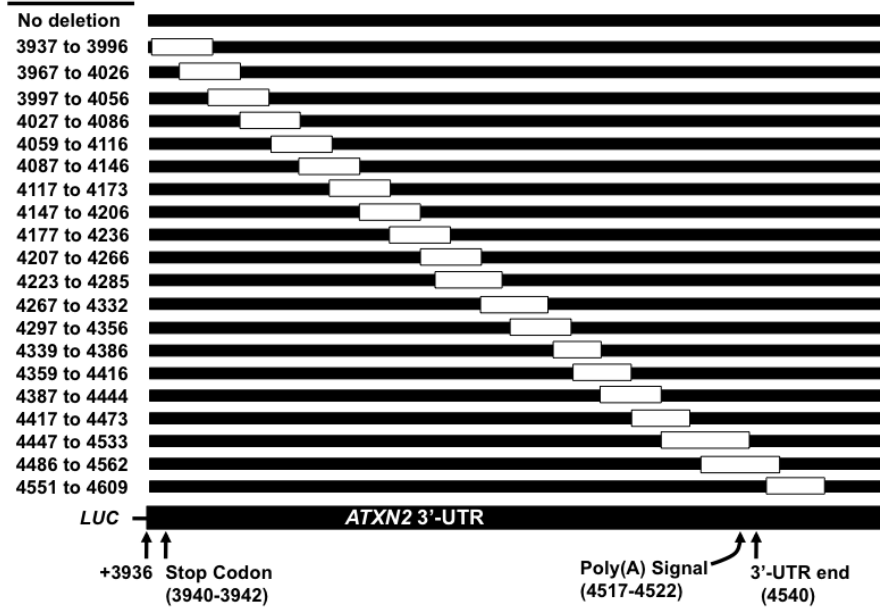
- Strong
- Moderate
- Weak
- Negative
- Not representative

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Annotation Summary - IHC

A

Region Deleted



B

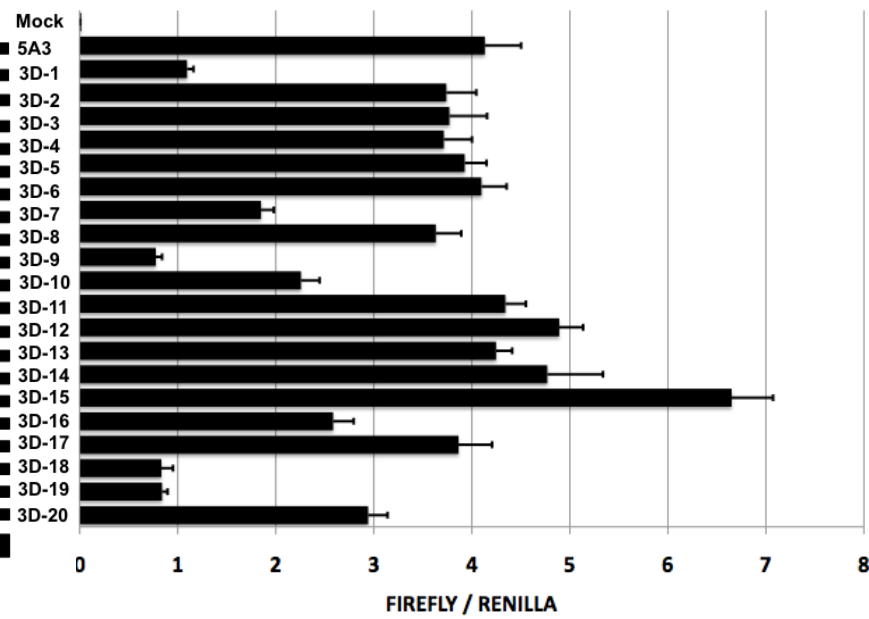


Fig 5

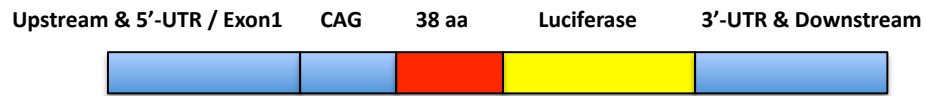
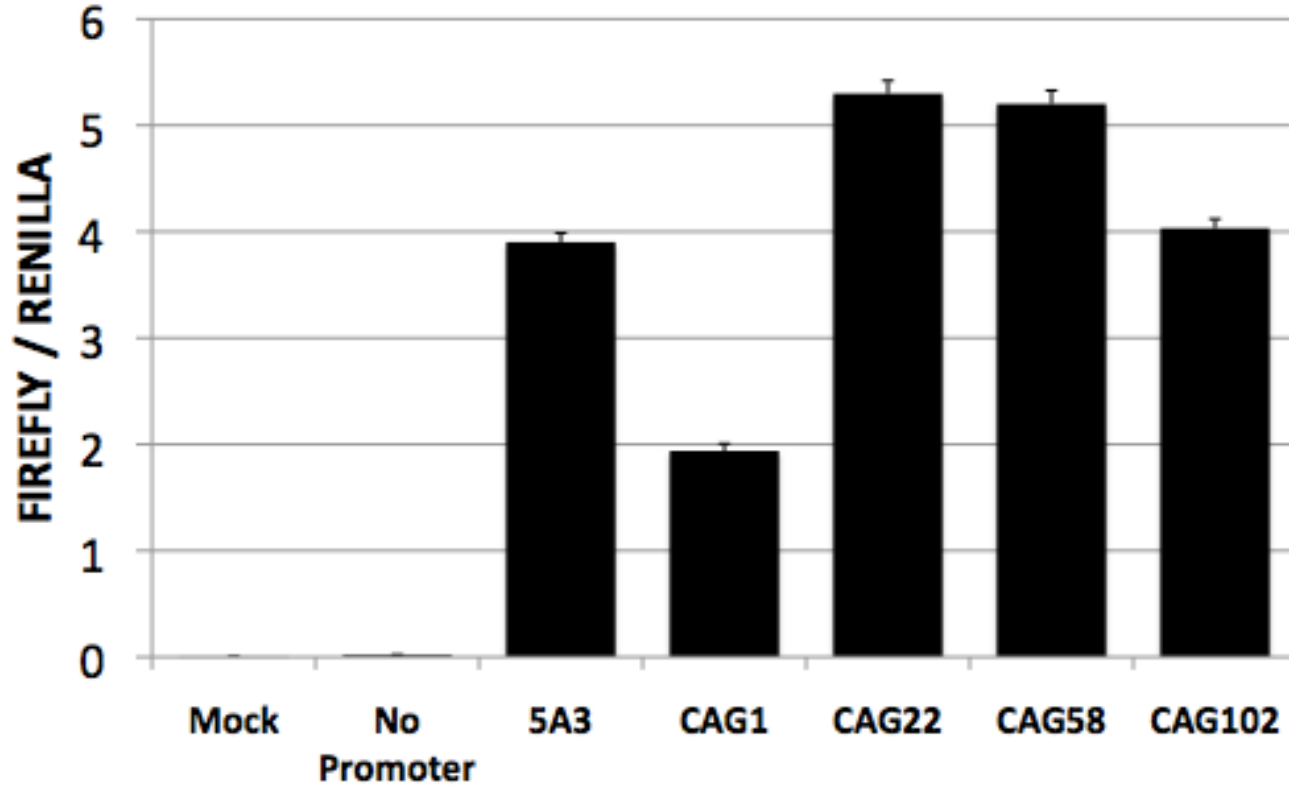


Fig 6

38 aa of ATXN2 following CAG repeat

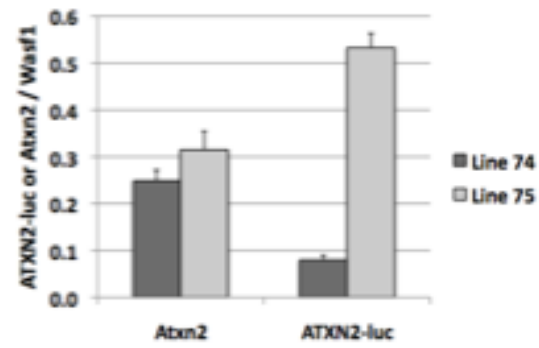
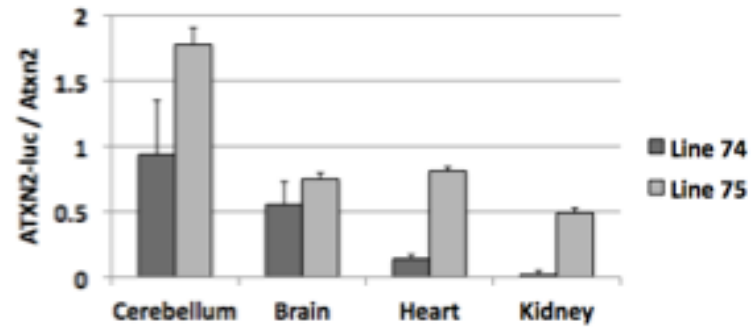
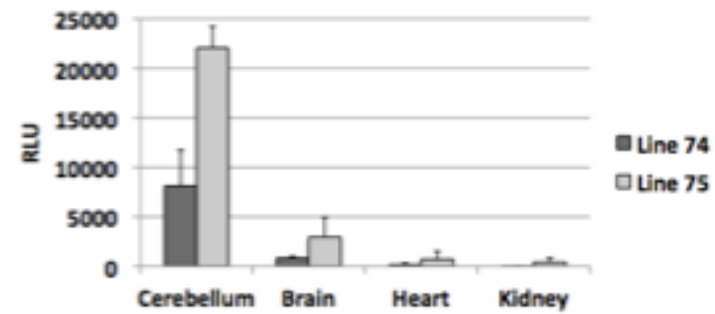
A**B****C**

Fig 7

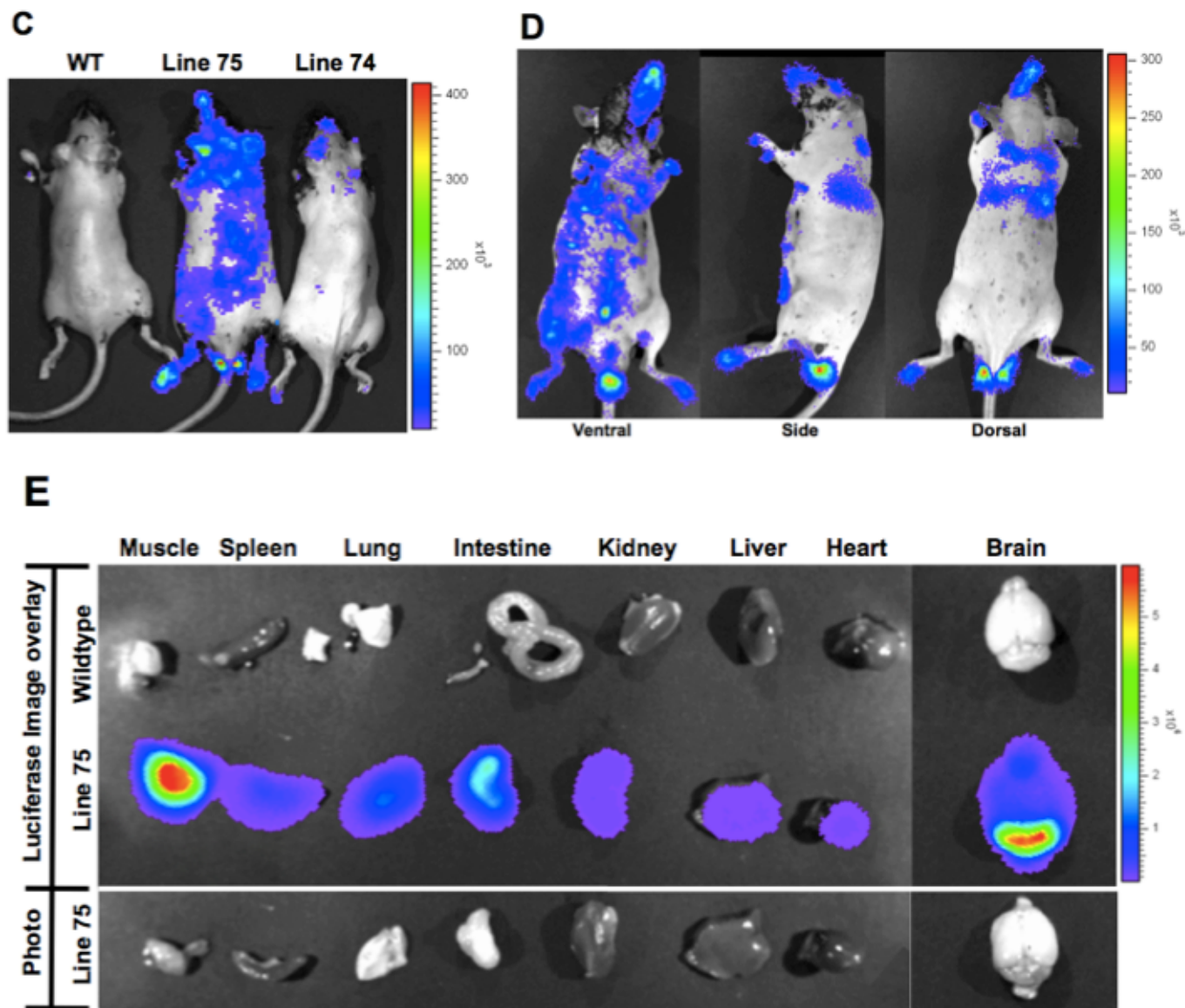


Fig 7

