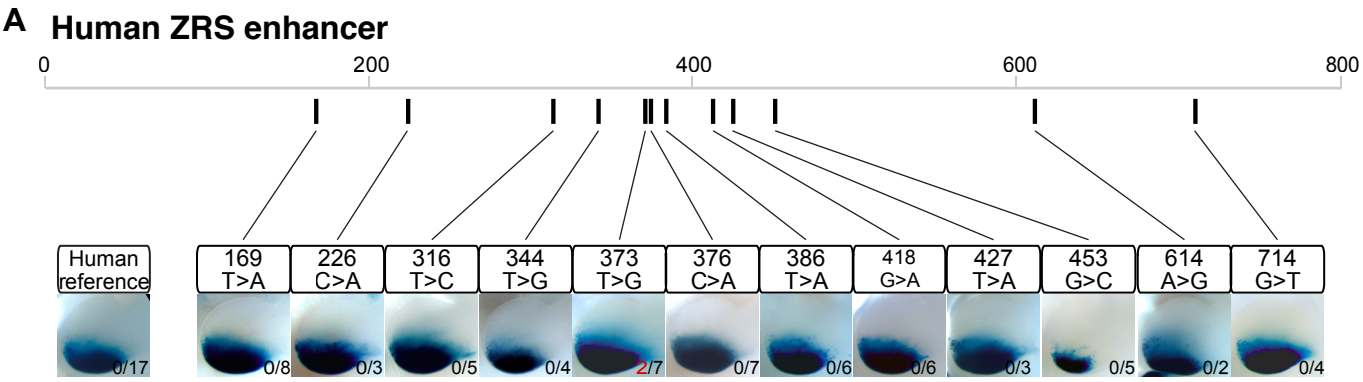

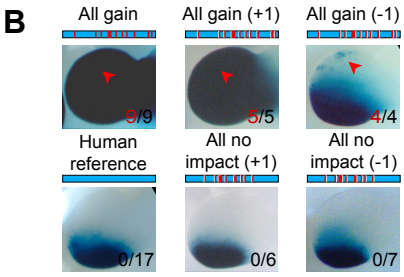

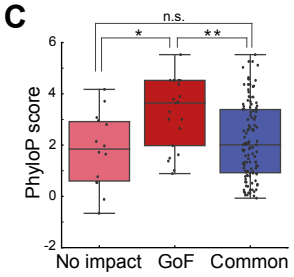

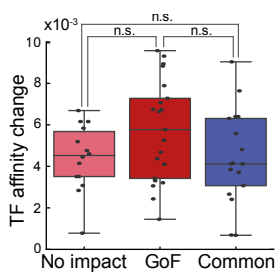

E

EXPRESSION GAIN (ANTERIOR) (18) | **NO IMPACT (11)**

Variant	406 A>G	739 A>G	621 C>G	619 C>T	743 T>G	407 T>A	417 A>G	404 G>A	404 G>T	404 G>C	401 A>C	401 A>G	287 C>A	463 T>G	402 C>T	105 C>G	396 C>T	334 T>G	329 T>C	574 G>A	507 C>G	428 T>A	71 C>G	328 C>G	305 A>T	297 G>A	295 T>C	165 A>G	41 A>C
Variant name	THA	USAC	USB	SAU	Aus	NA	FR3	CUB	IND	BRA	FR5	FR4	PAK1	PAK2	MEX	DU	TUR1	FR2	BEL2	NA	FR7	CHN	NA	IND2	BEL1	FR1	UKLP	DU2	NA
Phenotype severity	3	2	3	4	3	3	5	5	6	6	4	3	5	2	5	3	2	2	2	2	2	2	1	3	2	3	4	1	4
Has LOD score>3	NA	Yes	Yes	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	Yes	Yes	NA	NA	NA	NA	NA	NA	NA	NA	NA	Yes	NA	NA
>1 Pedigree	No	Yes	No	No	No	No	Yes	Yes	No	Yes	No	No	No	No	Yes	Yes	No	No	No	Yes	Yes	No	No	No	No	No	Yes	No	No
>1 Affected individuals	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	No	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	No	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Transmits	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	NA	Yes	Yes	Yes	Yes	Yes	Yes	Yes	NA	NA	NA	Yes	NA	Yes	Yes	Yes	Yes	Yes
Segregates	NA	Yes	Yes	Yes	NA	NA	NA	Yes	Yes	Yes	NA	NA	Yes	Yes	Yes	Yes	Yes	Yes	Yes	NA	NA	NA	Yes	NA	Yes	Yes	Yes	NA	NA
De novo	NA	NA	NA	NA	NA	NA	Yes	Yes/ NA	NA	NA	NA	Yes	NA	NA	NA	NA	NA	NA	NA	NA	NA	Yes	NA	NA	NA	NA	NA	NA	NA
Limb TFBS	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	No	Yes	No	Yes	Yes	Yes	No	No	Yes	Yes	Yes	Yes	Yes	Yes	Yes	No
Conserved (Human-fish)	Yes	No	No	Yes	No	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	No	Yes	Yes	No	Yes	No	Yes	Yes	Yes	Yes	Yes	Yes	Yes	No
Conserved (Human-mouse)	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	No	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Not present in cntrl group	Yes	Yes	Yes	Yes	Yes	NA	Yes	Yes	NA	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	NA	Yes	NA	Yes	Yes	NA	Yes	Yes	NA	Yes	Yes	NA
Not present in cntrl group >200	Yes	Yes	Yes	NA	NA	NA	Yes	Yes	NA	Yes	Yes	Yes	NA	Yes	NA	Yes	NA	NA	NA	NA	Yes	Yes	NA	Yes	NA	NA	Yes	NA	NA
Overlaps existing SNP	No	No	No	No	No	No	No	No	No	No	No	No	No	No	No	No	No	No	No	Yes	Yes	No	Yes	No	No	No	No	No	No
Full penetrance	Yes	No	Yes	Yes	No	Yes	Yes	Yes	Yes	Yes	Yes	NA	Yes	Yes	Yes	Yes	Yes	No	Yes	NA	Yes	NA	NA	NA	Yes	Yes	No	No	NA
Transgenic Activity																													