

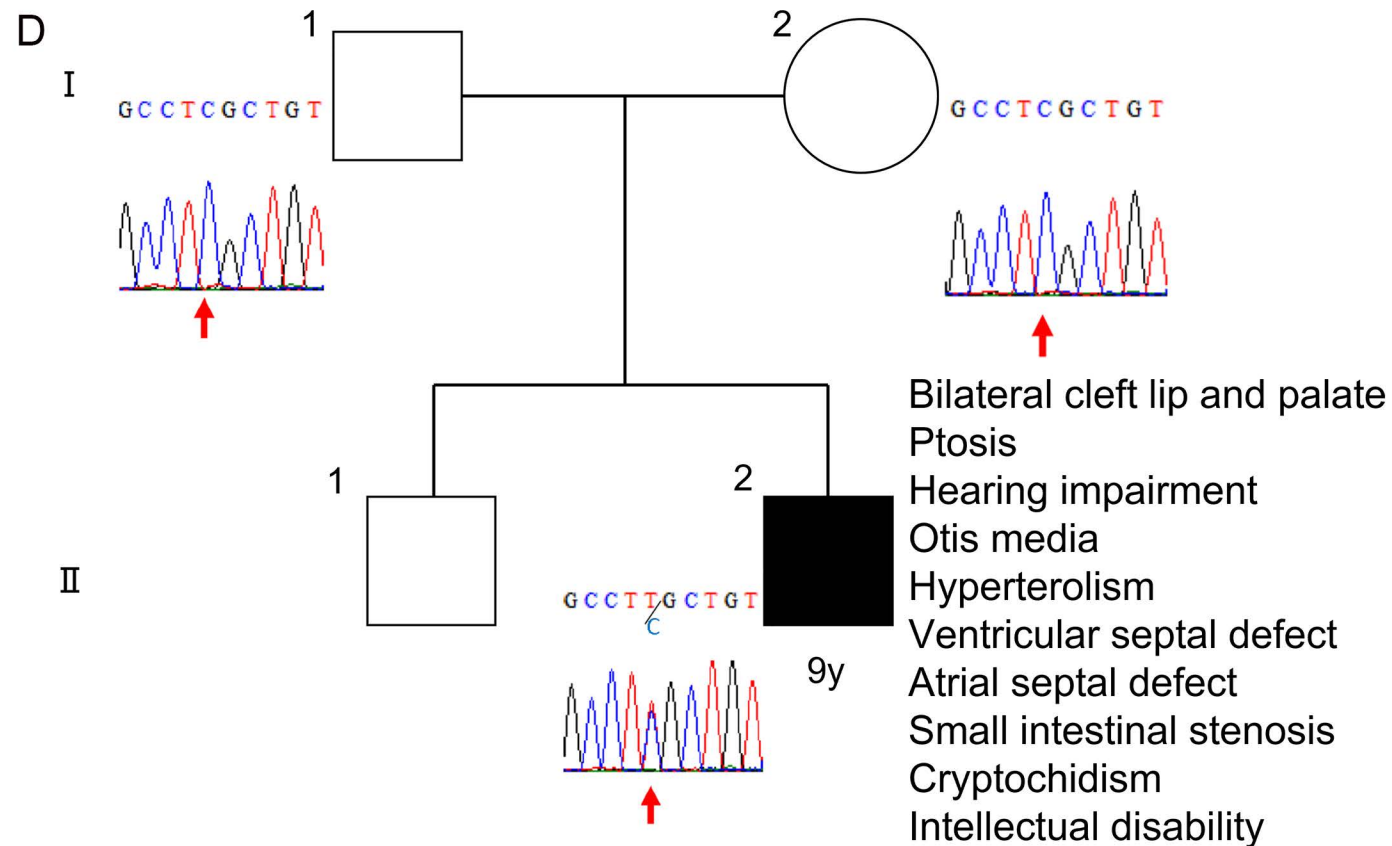
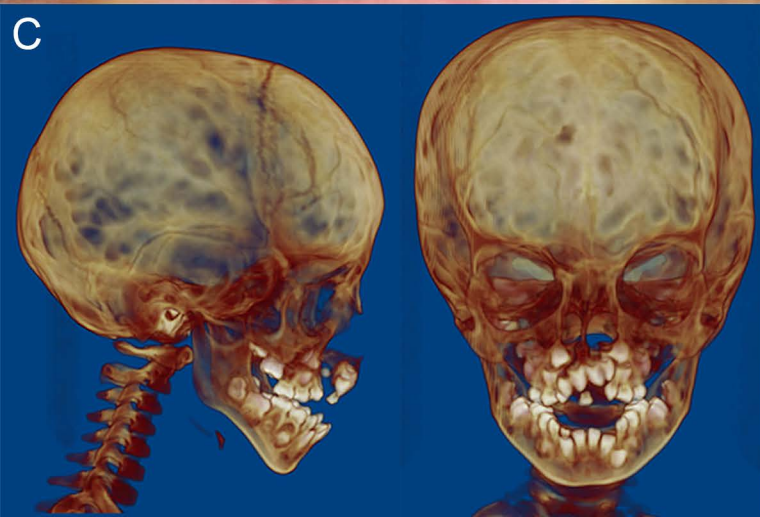


Title	Compromised actin dynamics underlie the orofacial cleft in Baraitser-Winter Cerebrofrontofacial syndrome with a variant in ACTB
Author(s)	Tsujimoto, Takayuki; Ou, Yushi; Suzuki, Makoto et al.
Citation	Human Molecular Genetics. 2024, 33(22), p. 1975-1985
Version Type	AM
URL	https://hdl.handle.net/11094/103591
rights	
Note	

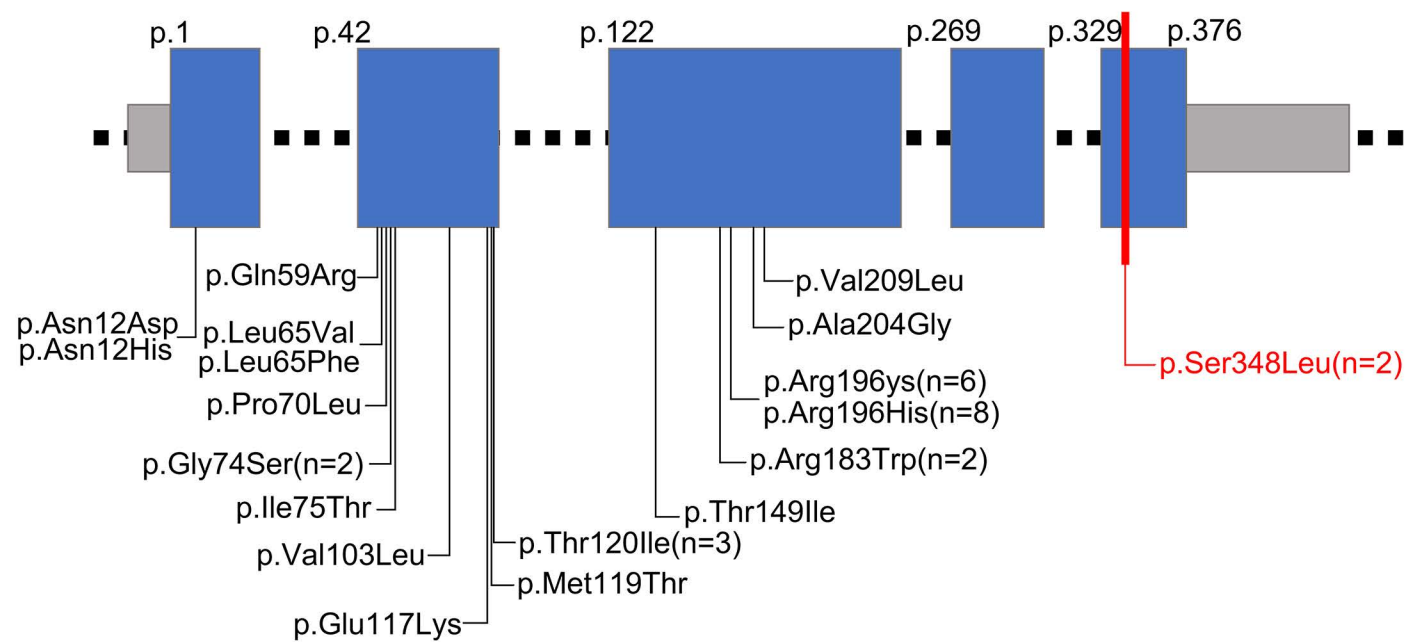
The University of Osaka Institutional Knowledge Archive : OUKA

<https://ir.library.osaka-u.ac.jp/>

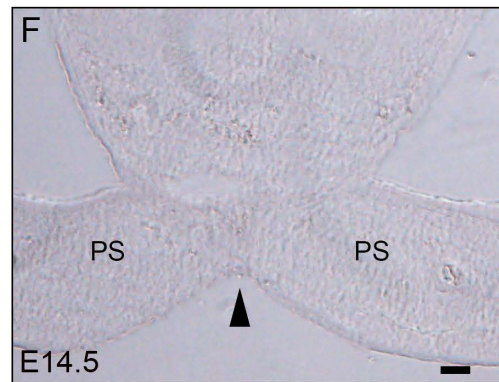
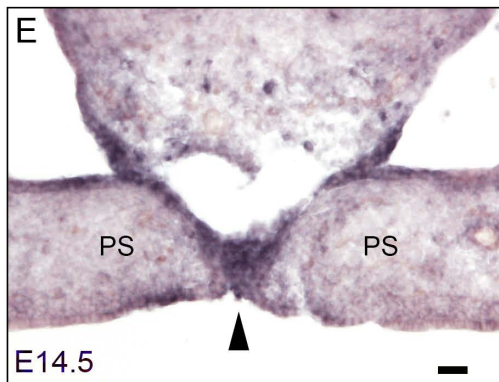
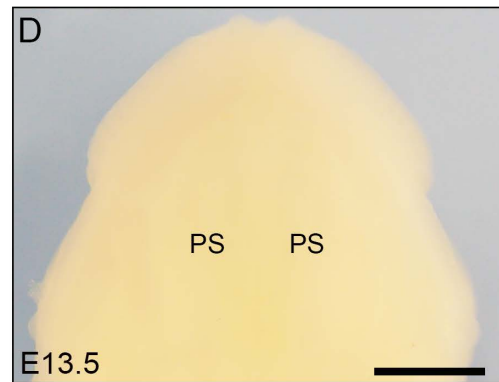
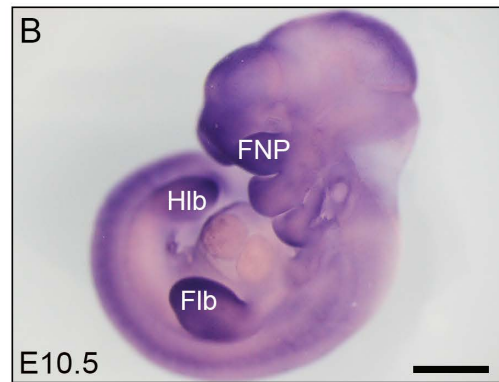
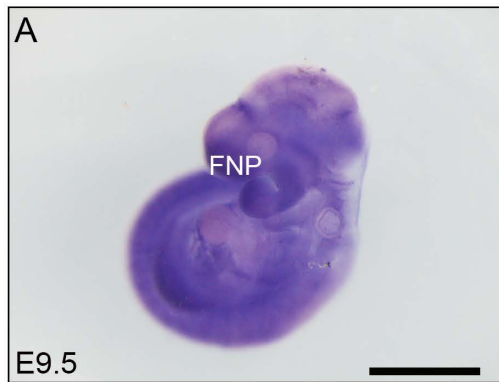
The University of Osaka



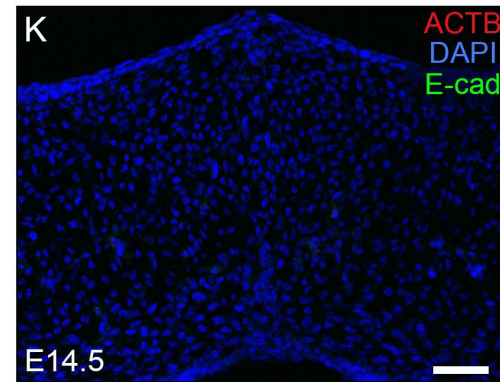
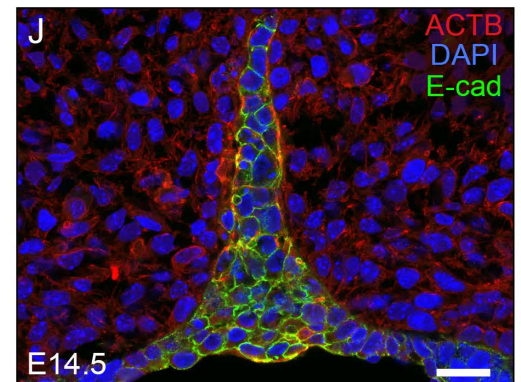
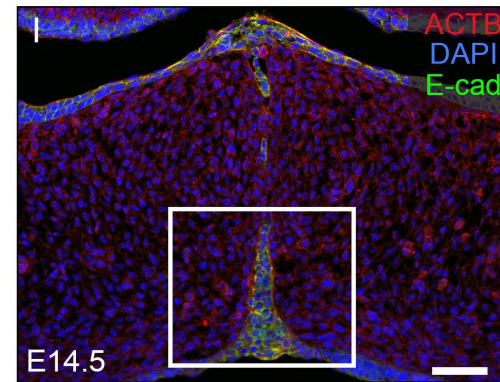
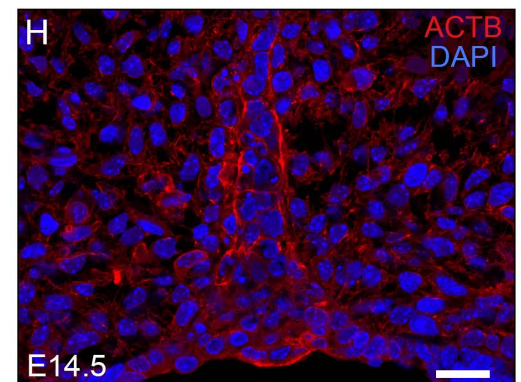
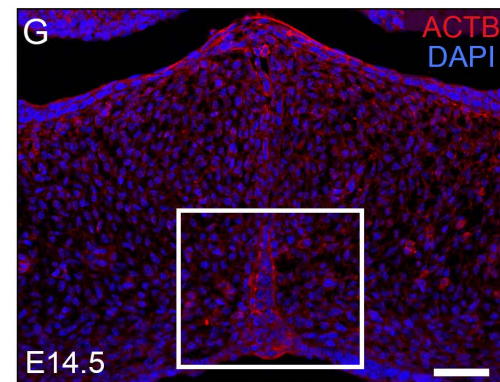
E *ACTB* gene mutations(chr7:5,566,779-5,570,232)

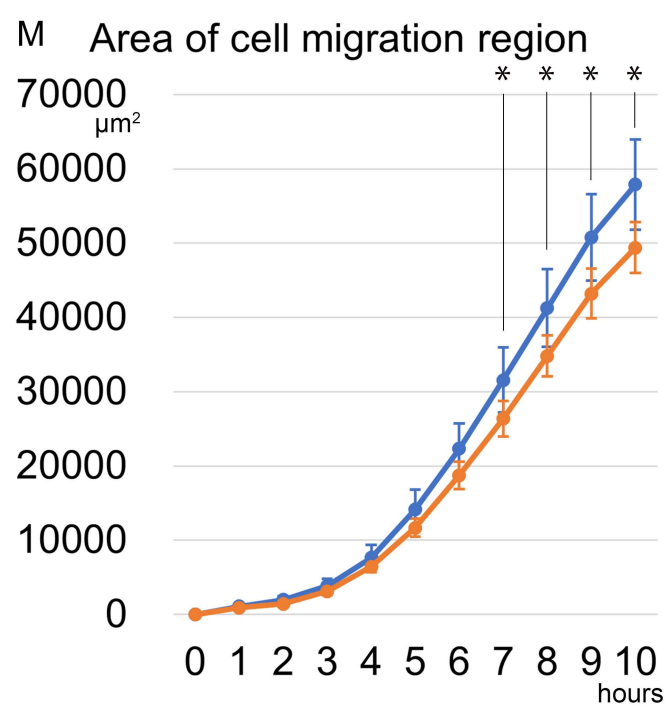
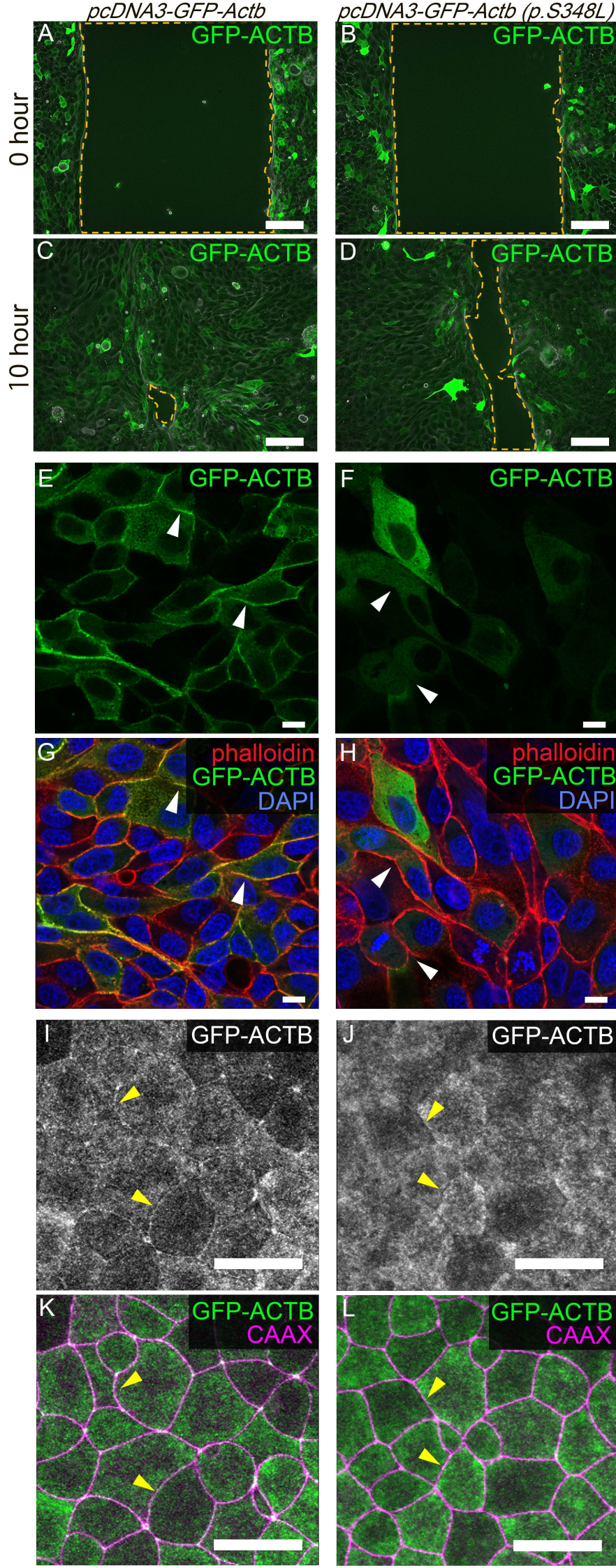


Actb mRNA



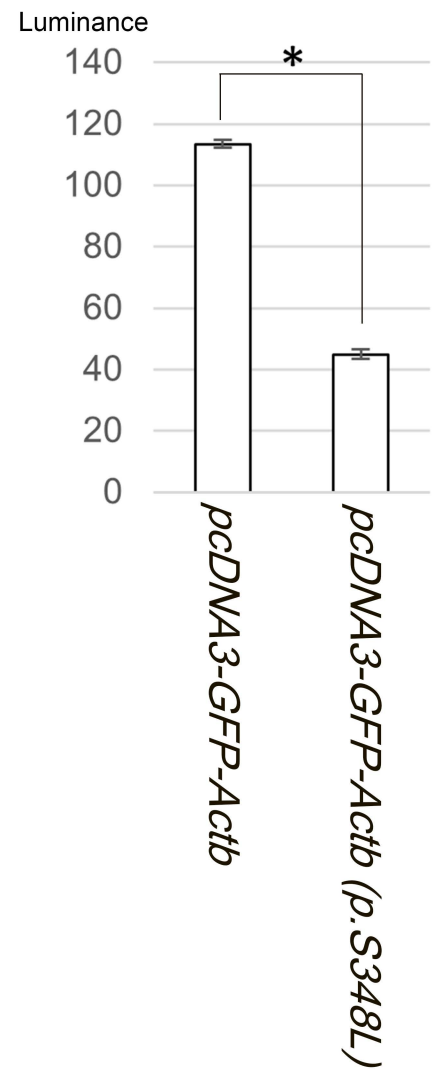
ACTB protein





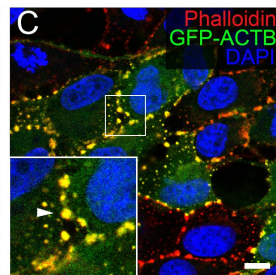
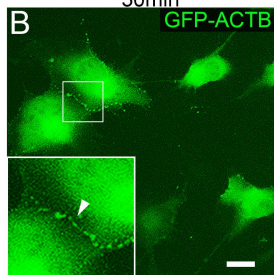
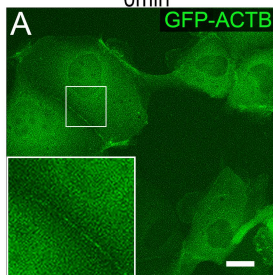
—●— *pcDNA3-GFP-ACTB*
 —●— *pcDNA3-GFP-ACTB(p.S348L)*

N Luminance at cell junction

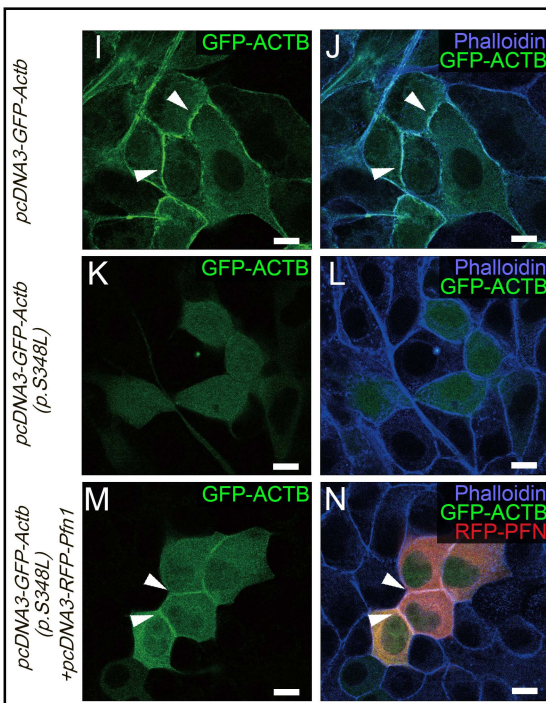
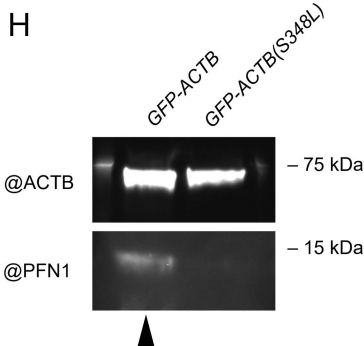
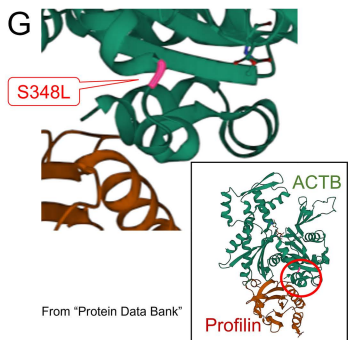
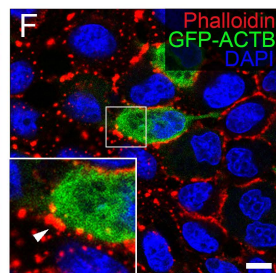
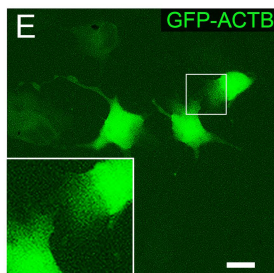
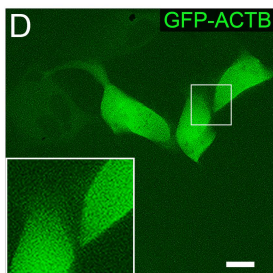


0min 30min

pcDNA3-GFP-Actb



pcDNA3-GFP-Actb
(p.S348L)



A

S348 DSB

CCATCTTGGCTTCGCTGTCCACCTTCCAGCAGATGTGG
GGTAGAACCGAAGGGACAGGTGGAAGGTCGTCCTACACC

gRNA target sequence

