

Supplemental Data

Homozygous Null *TBX4* Mutations Lead to Posterior

Amelia with Pelvic and Pulmonary Hypoplasia

Ariana Kariminejad, Emmanuelle Szenker-Ravi, Caroline Lekszas, Homa Tajsharghi, Ali-Reza Moslemi, Thomas Naert, Hong Thi Tran, Fatemeh Ahangari, Minoo Rajaei, Mojila Nasser, Thomas Haaf, Afrooz Azad, Andrea Superti-Furga, Reza Maroofian, Siavash Ghaderi-Sohi, Hossein Najmabadi, Mohammad Reza Abbaszadegan, Kris Vleminckx, Pooneh Nikuei, and Bruno Reversade

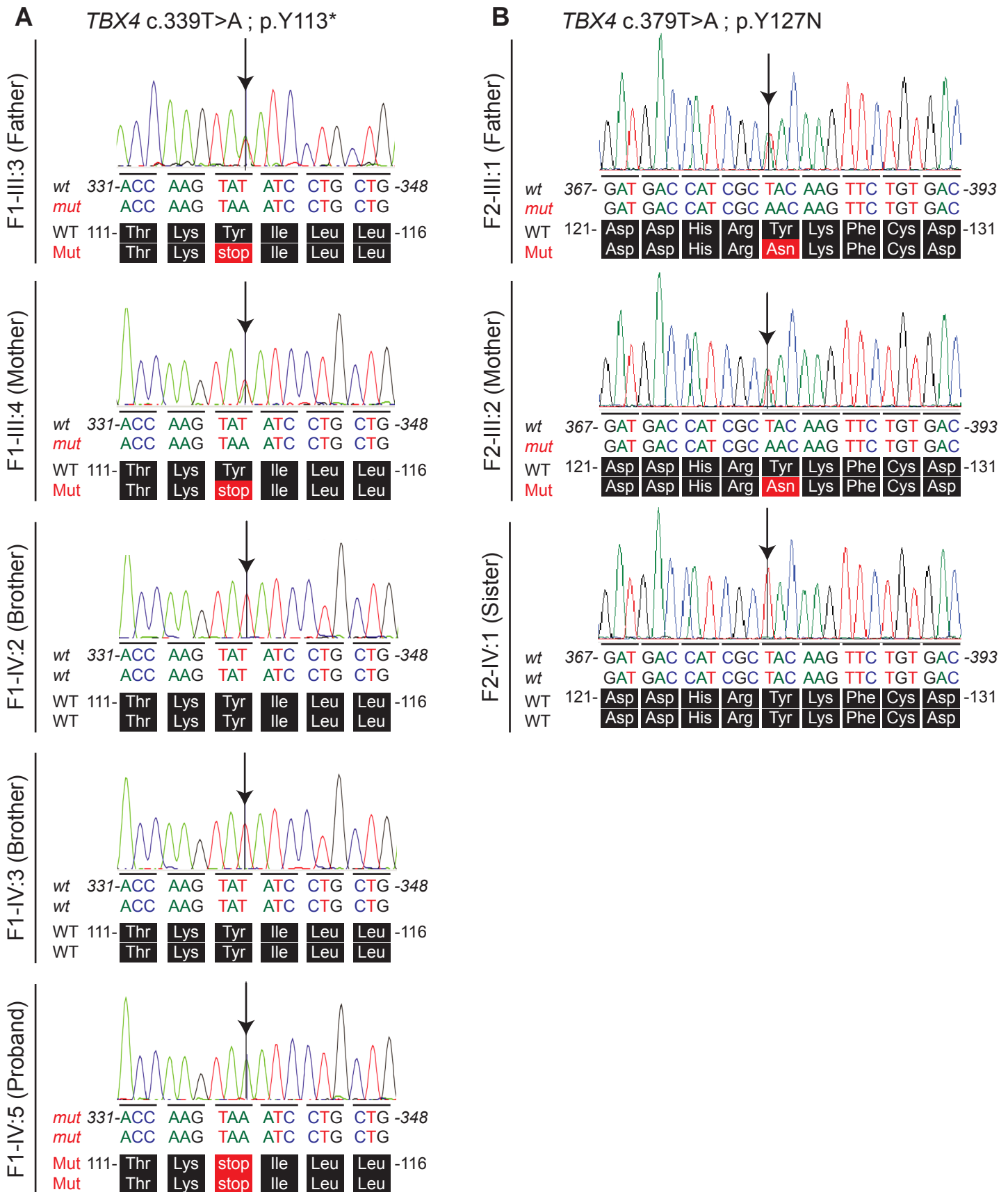


Figure S1: Segregation analysis of causative germline *TBX4* mutations. Sanger sequencing of *TBX4* mutations in available family members of Family 1 (a) and Family 2 (b). wt: wildtype, mut: mutant.

Tbx4 crispant

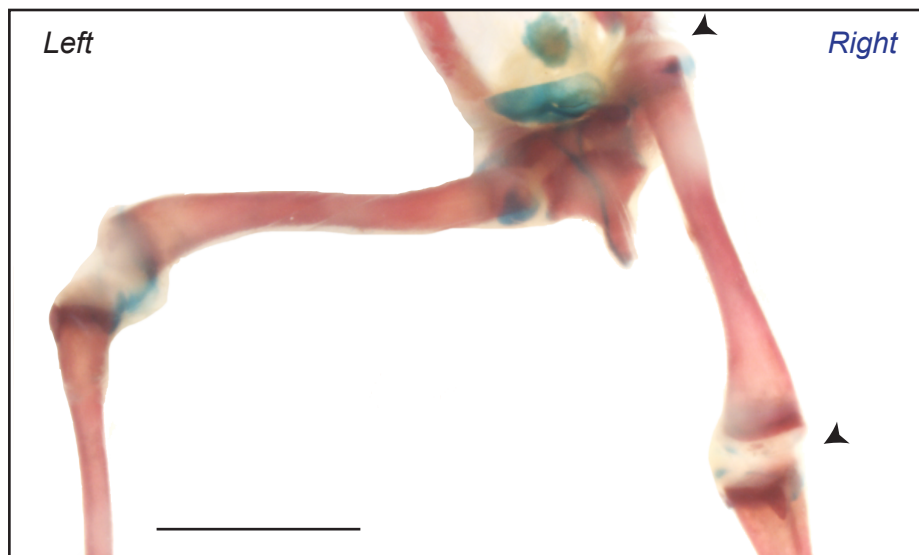
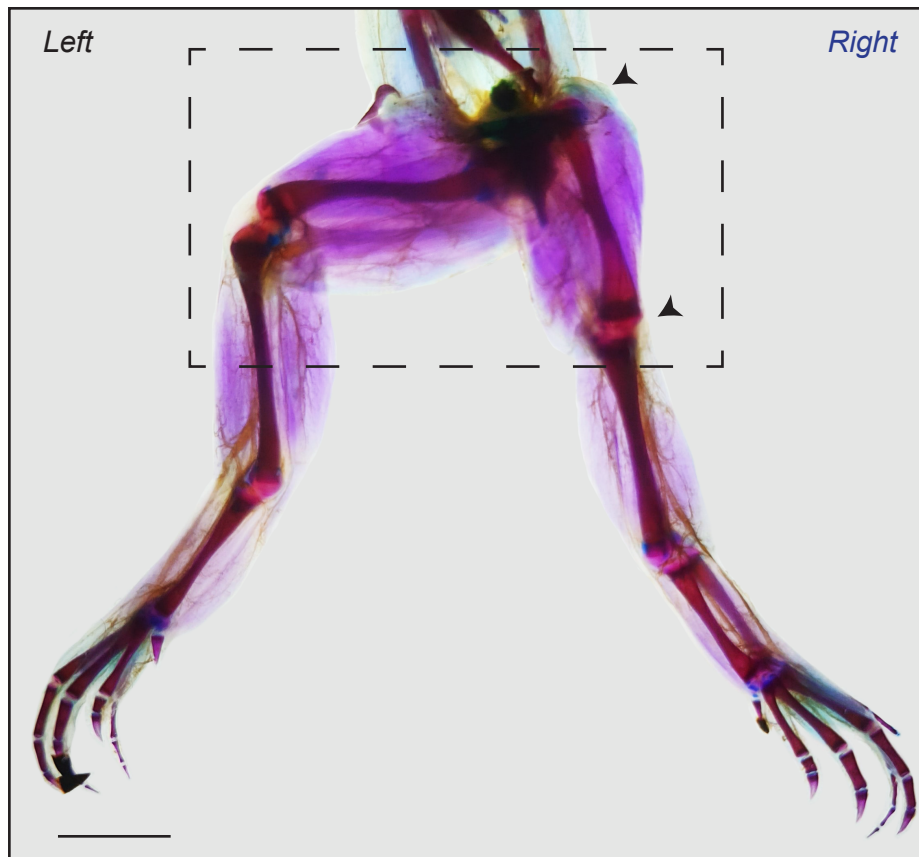


Figure S2: Phenotype of mildly affected *Xenopus tropicalis* animals. Representative images of an NF stage 66 adult animal, that had been unilaterally/ventrally injected with *Tbx4*_gRNA2 and Cas9 protein at the 4-cell stage, stained with alizarin red/alcian blue. Note the dislocated joints (black arrowheads) both at the hip and knee, and the shorter femur of the hindlimb in the injected side (right). Scale bars: 0.2 cm.