

Table S1. Summary of MYO7A allele and genotype distribution in the sub-Saharan South African DFNB2 families
Available family members_DNA $\Sigma n = 32$ Total number of individuals affected_Deaf $\Sigma n = 17$ Total number of variations = 8

MYO7A variation	Reference allele	Genotype	Family 1	Family 2	Family 3	Family 4	Family 5	Family 6	Family 7	Family 8	Family 9
			TS065/100 <i>n</i> = 5	TS074/093 <i>n</i> = 5	BS044 <i>n</i> = 9	TS076 <i>n</i> = 5	Shilo (TS074 branch C) <i>n</i> = 3	(TS074 branch B) <i>n</i> = 2	TS040 <i>n</i> = 1*	TS036 <i>n</i> = 1*	TS070 <i>n</i> = 1*
p.Tyr1780Ser	A	C/C	2				2	1			
		A/C	3	4		3	1	1			
p.Pro2126LeuTer5	TC	TC/T				5			1		
splice_region									1		
p.Ser617Pro	T	C/C			3						
		T/C			4						
p.Gly329Asp	G	G/A		4							
p.Arg373His	G	G/A								1	
p.Thr381Met	C	C/T								1	
p.Arg83Cys	C										1
Total affected (n)			2	3	3	3	2	1	1	1	1

Table S2. Alignment of the conserved second MyTH7 subdomain in different species and against p.Pro2126Leufs*5.

WT	CAA ACT ACG GAG CCA AAC TTC CCT GAG ATC CTC CTA ATT GCC ATC AAC AAG TAT GGG GTC AGC CTC ATC GAT CCC AAA
Translation +1	Q T T E P N F P E I L L I A I N K Y G V S L I A P L
Family TS076 MT	CAA ACT ACG GAG CCA AAC TTC CTG AGA TCC TCC TAA
	Q T T E P N F L R S S *

2114 FFEVKQTTEPNFLRS S*

p.Pro2126Leufs*5

HS mutant

2114 FFEVKQTTEPNFPE I LLIANKYGVSLID PKTKDILT THPFTKISNWSSGNTYFHI TIGNLVRGSKLLCETSLGYKMDDLTSYISQMLTAM

HS Wild type

2068 FFEVKQTTEPNYPE M LLIANKHGVSLIH PVTKDILV THPFTRISNWSSGNTYFHM TIGNLVRGSKLLCETSLGYKMDDLTSYISL MLTNM

DM

2064 FFEVKQTTEPNYPE M LLIANKHGVSLIH PSSKDILV THPFTRISNWSSGNTYFHM TIGNLVRGSKLLCETSLGYKMDDLTSYISL MLTNM

AA

2076 FFEVKQTTEPHFPE I LLIANKYGVSLID PKNKDILT TYPFTKISNWSSGNTYFHI TIGNLVQGSKLLCETSLGYKMDDLTSYISQMLTAM

DR

2114 FFEVKQTTEPNFPE I LLIANKYGVSLID PRTKDILT THPFTKISNWSSGNTYFHI TIGNLVRGSKLLCETSLGYKMDDLTSYISQMLTAM

MM

2076 FFEVKQTTEPNFPE I LLIANKYGVSLID PRTKDILT THPFTKISNWSSGNTYFHI TIGNLVRGSKLLCETSLGYKMDDLTSYISQMLTAM

SS

2068 FFEVKQTTEANYAE M LLIANKHGVSLIH PVTKDILV THPFTRISNWSSGNTYFHM TIGNLVRKLLCETSLGYKMDDLTSYISL MLTNM

DM mutant

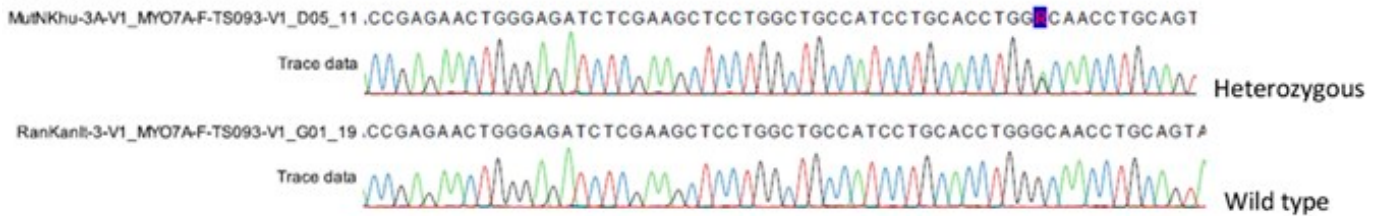
V1

V2

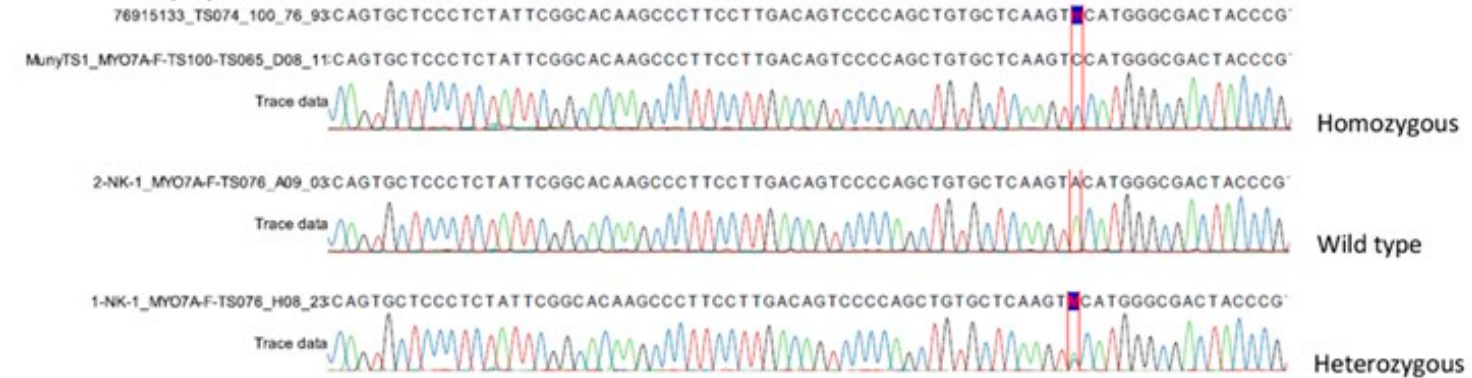
HS *Homo sapiens*; DM *Drosophila melanogaster*; AA *Anopheles arabienis*; DR *Danio rerio*; MM *Mus musculus*; SS *Sus scrofa*

Mutation p.Pro2126Leufs*5 results in a frameshift with termination of the protein after residue 5 downstream. The premature termination of the protein leads to interference with the conformation and regulation of the motor function of the motor domain. In their functional studies, Yang et al (2009) demonstrated that a point mutation induced at V2 abolished the folding of the tail region over the head domain and the autoregulation of the myosin 7a protein. Adapted from Yang et al., 2009.

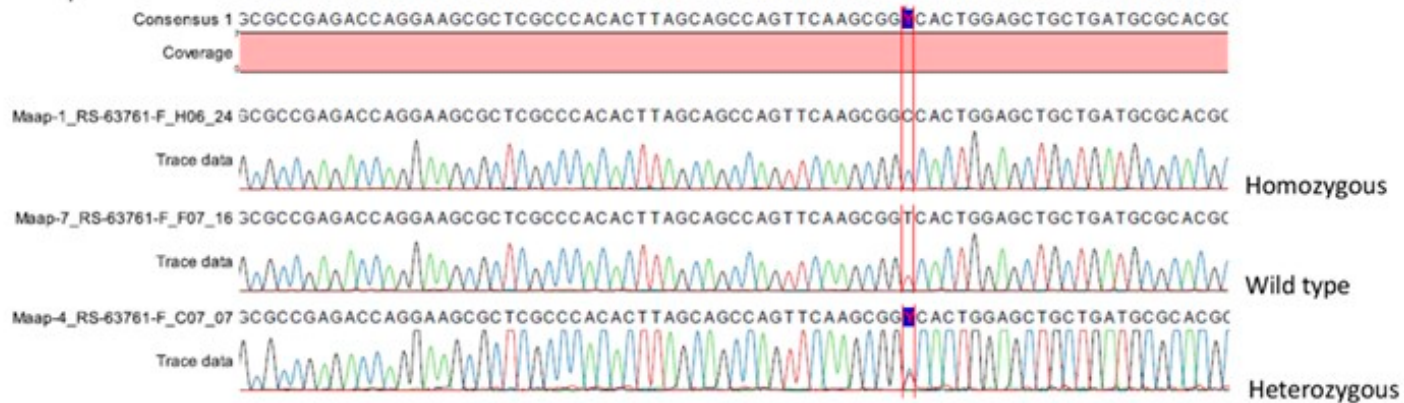
c.986G>A p.Gly329Asp



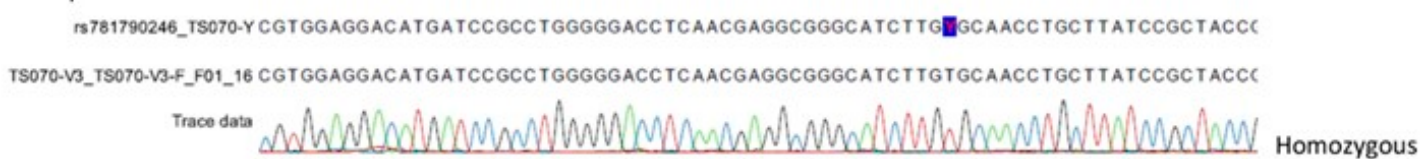
c.5339A>C p.Tyr1780Ser



c.1849T>C p.S617P



c.247C>T p.R83C



c.6375delC p.Pro2126Leu

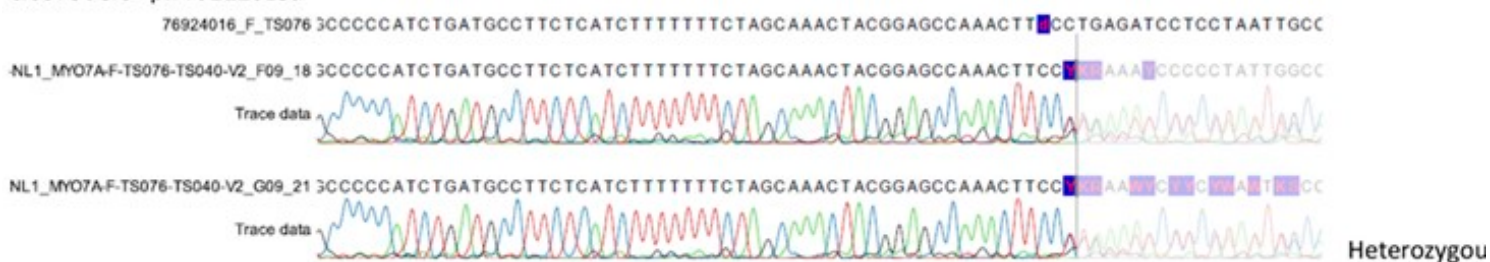


Figure S1: Sanger sequencing electropherograms of MYO7A mutations among South African DFN2 families.