

Alignment :

Dr-Sox17	MSSPDAGYSSDDPSQTSSCSSVMMPGMGQCPWVDPLSPLSDSKSKHEKCSAAGP-----G	55
Hs-SOX17	MSSPDAGYASDDQSQTSALPAVMAGLGPCPWAESLSPIGDMKVKGAPANS GAPAGAAG	60
Mm-Sox17	MSSPDAGYASDDQSQPRSAQPAVMAGLGPCPWAESLSPLGDVKVKGEVVASSGAPAGTSG	60
Dr-Sox17	RGKSEPRIRRP MNAFMVWAKDERKRLAQONPDLHNAELSKMLGKSWKALPMVDKRPFVVEE	115
Hs-SOX17	RAKGE SRIRRP MNAFMVWAKDERKRLAQONPDLHNAELSKMLGKSWKALTLAEKRPFVVEE	120
Mm-Sox17	RAKAE SRIRRP MNAFMVWAKDERKRLAQONPDLHNAELSKMLGKSWKALTLAEKRPFVVEE	120
Dr-Sox17	AERLRVKHMQDHPNFKYRPRRRKQVQRNKRLEPSFPLPGMCDAKM-TLCTEGMSAGYSGA	174
Hs-SOX17	AERLRVQHMQDHPNFKYRPRRRKQVQRNKRLEGGF-LHGLAEPQAAALGPEGGRVAMDGL	179
Mm-Sox17	AERLRVQHMQDHPNFKYRPRRRKQVQRNKRLEGGF-LHALVEPQAGALGPEGGRVAMDGL	179
Dr-Sox17	GLPQYCENHTLFESYSLPTDPSMDAGTTEFFAQLQDQSAFSYHHQOEHHFQEQTNILN	234
Hs-SOX17	GLQFPEQGFAGPPLLPPHMGGHYRDCQSLGAPPLDGYPLPTPDTSPLDGVDPDPAFFAA	239
Mm-Sox17	GLPFPEPGYPAGPPLMSPHMGPHYRDCQGLGAPALDGYPLPTPDTSPLDGVEQDPAFFAA	239
Dr-Sox17	DTHCHGNTQTLKSRQSHSIAYSNINTNTNSNLHAPINAQLSSINLQQVFHENANPQISHH	294
Hs-SOX17	PMPGDCPAAGTYSYAQVSDIAGPEPPAGPMHPRLGPEPAGPSIPGLLAPPSALHLYYGA	299
Mm-Sox17	PLPGDCPAAGTYTYAPVSDYAVSVIEPPAGPM--RVGPDPSGPAMPGILAPPSALHLYYGA	297
Dr-Sox17	PGTHLNIFNRSPSSSSHAMTFA---YLNCPSTLDTFYNSSSQMKELSHCVSSHTHKQQS	351
Hs-SOX17	MGSPGAGGGRGFQMPQHQQHQ-----HQHHP-GPGQSPPEALPCRDGTDPSQPA	352
Mm-Sox17	MGSPAASAGRGFHAQQQPLQPQAPPPPPQQQHPAHGPGQSPPEALPCRDGTESNQPT	357
Dr-Sox17	IAEAQSQASTATHSSGMVDEVEFEHCLSFVGSAPLPGSDLISTVLSDASSAVYICGYN	411
Hs-SOX17	ELLGEVDRTEFEQYLHFVCKPEMGLPYQGHDSGVNLPDSHGAISSVSDASSAVYICNYP	412
Mm-Sox17	ELLGEVDRTEFEQYLPFVYKPEMGLPYQGHDCGVNLSDSHGAISSVSDASSAVYICNYP	417
Dr-Sox17	NS	413
Hs-SOX17	DV	414
Mm-Sox17	DI	419

Supp. Figure S1. Alignment of vertebrate Sox17 protein sequences. Residues identical in all species are highlighted in yellow, those conserved only in two species are highlighted in blue. Tyrosine residues (Y) predicted to undergo phosphorylation are highlighted in red. The HMG box is indicated by a black line, the G/P-rich region by a red line. Red arrows indicated the location of the mutations. The Accession numbers for the sequences are as follows: Dr-Sox17 (NP_571362), Hs-SOX17 (NP_071899), Mm Sox17 (NP_035571). Dr: *Danio rerio*, Hs: *Homo sapiens*, Mm: *Mus musculus*.

Supp. Table S1. Molecular characterization of the duplicated chromosome 8q region in Family 1

Marker	Position (Kb)	Proband	Mother	Father	der(8)	chr. 8	Results
D8S1750	35470	216/220	216/218	216/220	ND	ND	N
D8S1821	38369	146/164	140/164	146	ND	ND	N
D8S268	41264	255/257	257	255/257	ND	ND	N
D8S1115	42544	164	161/164	161/164	ND	ND	U
Centromere							
D8S531	49074	117*/119	115/117	115/119	117	119	dup(M)
D8S601	53846	219/227*	223/227	219/231	227	219	dup(M)
D8S1737	54949	191	191	191	191	191	U
SOX17 E2 T/A	55534	T/A*	T/A	T	A	T	dup(M)
D8S509	55756	269*/271	269	271	269	271	dup(M)
D8S1828	56962	205	205	205	205	205	U
D8S260	60000	208/210	210	208	210	208	N

The proband, her mother and father, and two somatic cell hybrid clones containing either the duplicated chromosome 8 (der(8)) or the normal chromosome 8 (chr. 8), were assayed. Duplicated alleles are indicated by an asterisk (*). N, normal chromosomal asset; U, uninformative; dup(M), duplication of maternal origin; ND, not done.