

**Supplementary Table 1** Results of quantitative PCR analyses for the human TES region for 66 XY gonadal dysgenesis cases (Freiburg cohort A, 44 cases; Münster cohort B, 22 cases).

Case	Amplicon			
	5' TESCO	TESCO-A	TESCO-B	3' TESCO
L1734 <sup>a</sup>	0.46	0.43	0.47	0.43
Control <sup>b</sup>	1.02	1.01	0.95	0.97
A-1	0.82	1.02	1.09	1.28
A-2	0.90	1.00	0.94	1.12
A-3	0.87	1.08	0.89	1.03
A-4	0.96	1.13	1.05	0.90
A-5	1.03	0.86	1.01	1.15
A-6	0.96	0.95	0.82	1.06
A-7	1.03	0.92	1.01	1.12
A-8	1.12	0.90	1.13	1.16
A-9	0.95	0.85	1.00	1.05
A-10	0.90	0.99	1.14	1.00
A-11	1.05	0.88	0.83	1.06
A-12	1.02	0.98	1.15	0.98
A-13	1.10	1.10	1.14	1.26
A-14	0.97	1.01	1.00	1.08
A-15	0.99	0.88	1.06	1.01
A-16	0.92	-	0.86	1.08
A-17	1.01	1.01	1.02	0.99
A-18	1.09	1.07	1.07	1.16
A-19	0.93	0.87	0.87	1.05
A-20	0.91	0.91	0.99	0.99
A-21	1.01	0.94	1.06	1.09
A-22	0.92	0.93	0.98	1.03
A-23	0.78	1.03	1.11	0.91
A-24	1.02	1.06	1.04	1.07
A-25	0.97	0.94	1.04	1.04
A-26	0.93	0.98	1.05	1.01
A-27	1.10	0.99	1.03	1.04
A-28	1.07	1.12	1.03	1.14
A-29	1.10	1.05	1.03	1.06
A-30	0.93	0.96	0.94	1.16
A-31	0.98	0.92	1.06	1.15
A-32	0.93	0.93	0.88	1.11
A-33	1.03	0.97	0.95	0.87
A-34	0.87	0.98	1.06	0.83
A-35	0.95	1.01	0.96	0.83
A-36	1.10	1.05	1.07	1.11
A-37	0.94	1.04	1.06	0.89
A-38	1.12	1.06	1.07	1.04
A-39	1.02	1.01	0.95	0.97
A-40	0.95	0.95	0.98	0.82

A-41	1.08	1.07	1.12	0.95
A-42	0.95	0.98	0.99	0.77
A-43	0.89	1.03	1.05	0.90
A-44	0.98	1.04	0.88	0.83

Case	Amplicon			
	5' TESCO	TESCO-A	TESCO-B	3' TESCO
L1734 <sup>a</sup>	0.46	0.43	0.47	0.43
Control <sup>b</sup>	1.02	1.01	0.95	0.97
B-1	1.08	1.08	0.98	1.08
B-2	1.10	1.11	0.89	1.18
B-3	1.08	0.86	0.94	1.07
B-4	0.97	0.97	1.09	1.13
B-5	1.03	1.04	1.04	1.08
B-6	1.03	0.86	0.94	1.07
B-7	1.08	0.89	1.04	1.15
B-8	1.10	1.07	0.96	1.02
B-9	1.09	0.90	1.01	1.09
B-10	1.17	0.90	1.05	0.95
B-11	1.11	1.13	1.03	1.04
B-12	1.03	0.97	1.17	0.97
B-13	0.96	0.91	0.91	1.13
B-14	0.93	1.00	0.83	0.86
B-15	1.04	0.94	0.97	1.02
B-16	1.20	1.08	1.11	1.15
B-17	1.08	0.94	1.09	1.13
B-18	1.17	1.04	1.10	1.18
B-19	1.03	1.00	0.98	1.07
B-20	1.23	1.02	1.00	1.12
B-21	1.22	1.07	1.01	1.14
B-22	1.04	1.04	1.03	1.09

The means of normalized ratios (mean of test amplicon values / mean average of both reference amplicon values) shown are averages of at least two measurements, each done in duplicate.

<sup>a</sup>*SOX9* heterozygous deletion case spanning the TES region, patient 1 in Pop et al. (2004)

<sup>b</sup>Wildtype control

-, failed PCR due to poor DNA quality