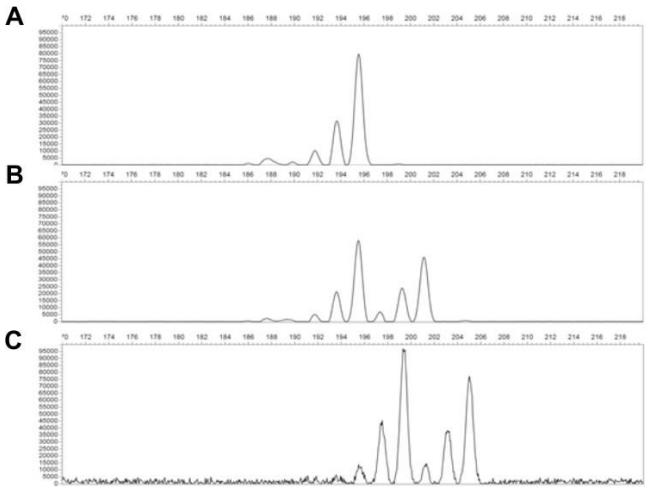
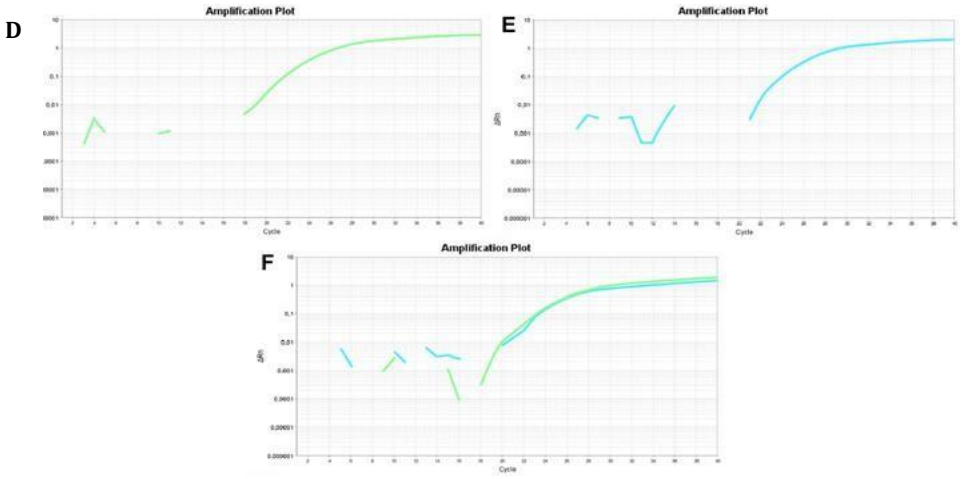
SSupplemental material

22q11.2 deletion syndrome: evidence of no influence of parental origin on clinical heterogeneity

Graphic result examples for microsatellite DNA markers and SNPs of one of the families (patient, mother, and father) is showed below.



Supplemental Figure S1. (**A**) Graphic visualization of the DNA microsatellite genotyping of a proband, which due to the deletion presents only one allele, 196bp in size. (**B** and **C**) Alleles of the mother and father respectively, both heterozygous, one of the mother's alleles (196bp) has the same fragment size as the allele present in the proband (196bp), indicating that the transmitted allele to the proband comes from the mother, revealing a paternal origin of the deletion.



Supplemental Figure S2: (**D**) Proband's graph of allele curves obtained with qPCR, showing only one green curve, due to the deletion, determining that the proband has a C allele. (**E**) Father's graph that displays only one blue curve, indicating T/T homozygosity. (**F**) Mother's graph shows both blue and green curves, indicating C/T heterozygosity. Since only the proband and the mother exhibit the green curve, it can be inferred that the allele transmitted to the proband is of maternal origin, revealing a paternal origin of the deletion.

Supplemental Table S1: Parental origin definition by genotyping of DNA microsatellite markers and SNPs.

Family	**D22S1638**	**D22S941**	**D22S944**	**D22S1623**	**D22S264**	**rs4819519**	**rs5993650**	**Origin**
P1	2	**8**	**4**	6	**3**	C	C	MAT
M1	7/7	13/13	5/5	6/6	2/9	C/T	C/T	
F1	-	**8/16**	**4/7**	6/6	**3/9**	C/T	C/T	
P2	**4**	**13**	5	6	**3**	-	-	MAT
M2	5/9	8/8	5/9	6/6	7/9	-	-	
F2	**4/9**	**8/13**	5/5	6/6	**3/3**	-	-	
P3	**8**	8	**6**	5	**7**	**C**	C	MAT
M3	5/7	8/8	4/5	5/6	8/9	T/T	C/C	
F3	**7/8**	8/13	**5/6**	5/5	**7/10**	**C/T**	C/T	
P4	5	**13**	**4**	**6**	**7**	C	T	MAT
M4	-	4/8	6/7	2/5	6/9	C/T	C/T	
F4	-	**13/13**	**4/5**	**5/6**	**7/10**	C/T	T/T	
P5	7	**3**	**6**	-	7	C	**T**	MAT
M5	6/7	8/16	5/7	5/6	6/7	C/T	C/C	
F5	4/7	**3/8**	**5/6**	5/6	6/7	C/T	**C/T**	
P6	**8**	**3**	**4**	5	**10**	C	**T**	PAT
M6	**5/8**	**3/13**	**4/5**	5/6	**7/10**	C/T	**T/T**	
F6	2/5	8/14	5/6	1/5	3/6	C/T	C	
P7	**5**	**13**	**5**	**5**	**7**	**C**	**T**	PAT
M7	**2/5**	**13/15**	**4/5**	**5/6**	**7/8**	**C/C**	**C/T**	

P8	**6**	4	**3**	6	**8**	-	-	MAT
M8	7	4/14	4/5	1/6	6/7	-	-	
F8	**4/6**	8/12	**3/5**	5/6	**7/8**	-	-	
P9	7	8	**8**	**7**	8	C	C	MAT
M9	7/9	8/8	4/7	5/5	8/9	C/T	C/T	
F9	5/7	8/14	**4/8**	**5/7**	6/8	C/C	C/C	
P10	**7**	8	**2**	6	9	C	C	MAT
M10	5/8	8/12	5/7	6/8	9/9	C/C	C/C	
F10	**5/7**	8/9	**2/7**	5/6	2/9	C/T	C/C	
P11	**4**	12	**6**	5	**7**	-	T	MAT
M11	3/7	12/16	4/4	5/5	2/9	C/C	C/T	
F11	**3/4**	8/12	**4/6**	5/5	**7/7**	T/T	C/T	
P12	3	**3**	5	5/5	9	T	**T**	MAT
M12	3/2	8/14	5/6	5/5	7/9	T/T	C/C	
F12	3/2	**3/8**	4/5	5/6	9/11	C/T	**C/T**	
P13	**2**	**7**	5	**6**	**8**	T	C	PAT
M13	**2/13**	**7/12**	5/5	**6/6**	**7/8**	C/T	C/T	
F13	3/5	4/13	5/6	3/4	6/9	C/T	C/C	
P14	7	**11**	4	**6**	**8**	C	T	MAT
M14	7/7	12/15	4/5	4/4	2/7	C/C	C/C	
F14	6/7	**7/11**	4/5	**6/6**	**8/10**	C/C	-	
P15	**6**	**7**	**4**	**4**	**8**	C	**C**	MAT
M15	2/7	4/12	5/5	6/6	6/10	C/C	T/T	
F15	**5/6**	**7/7**	**4/4**	**4/6**	**7/8**	C/C	**C/C**	
P16	**6**	12	5	6	**2**	C	**C**	PAT
M16	**6/6**	7/7	5/5	6/9	**2/8**	C/T	**C/C**	
F16	5/7	2/12	5/10	3/6	8/8	C/C	T/T	
P17	4	7	**5**	6	**2**	**C**	**T**	PAT
M17	2/4	7/12	**5/11**	4/6	**2/11**	**C/C**	**C/T**	
F17	4/6	7/11	6/7	5/6	8/9	T/T	C/C	
P18	**2**	12	**4**	**1**	**10**	T	**T**	PAT
M18	**2/7**	12/13	**4/5**	**1/6**	**2/10**	T/T	**T/T**	
F18	5/7	5/12	5/5	4/6	7/7	T/T	C/C	
P19	7	**12**	**5**	**4**	**7**	-	-	PAT
M19	7/7	**12/13**	**5/5**	**4/4**	**7/10**	-	-	
F19	7/8	8/15	3/6	6/8	9/12	-	-	
P20	**2**	7	**6**	**4**	7	C	C	PAT
M20	**2/2**	7/12	**6/10**	**4/6**	2/7	C/T	C/T	
F20	6/6	7/12	5/5	6/6	7/9	C/C	C/T	
P21	9	**13**	**5**	5	**1**	**T**	T	PAT
M21	9/9	**7/13**	**5/5**	5/5	**1/1**	**C/T**	C/C	
F21	3/9	7/12	4/4	5/5	8/8	-	-	
P22	9	12	**5**	**4**	**1**	-	-	PAT

M22	9/9	12/12	**3/5**	**4/4**	**1/7**	-	-	
F22	9/9	12/15	3/9	5/5	8/8	C/T	C/T	
P23	9	**14**	**8**	**5**	**9**	**T**	C	PAT
M23	9/9	**2/14**	**3/8**	**4/5**	**5/9**	**C/T**	C/T	
F23	6/9	7/12	6/10	4/4	1/10	C/C	C/T	
P24	**5**	**13**	**5**	5	**11**	C	**T**	MAT
M24	3/9	7/9	3/6	4/5	1/6	C/T	C/C	
F24	**5/5**	**13/13**	**4/5**	5/5	**9/11**	C/T	**T/T**	
P25	**5**	**6**	**5**	3	11	-	C	MAT
M25	**9**	**5/8**	**3/3**	2/4	5/10	C/T	C/C	
P26	**3**	**15**	**3**	**5**	**9**	C	C	MAT
M26	9/9	2/4	4/4	4/4	4/6	C/C	C/T	
F26	**1/3**	**15/15**	**3/4**	**5/5**	**8/9**	-	-	
P27	9	**12**	**4**	4	**8**	-	-	PAT
M27	7/9	**8/12**	**4/4**	4/4	**1/8**	C/T	C/T	
F27	6/9	2/8	5/5	4/4	1/5	C/T	C/T	
P28	**5**	**13**	**5**	**5**	**10**	C	**T**	PAT
M28	**4/5**	**13/13**	**5/5**	**5/6**	**9/10**	C/T	**T/T**	
F28	1/1	7/7	4/4	4	7/8	C/C	C/C	
P29	4	**4**	5	**5**	**6**	C	T	MAT
M29	4/4	13/13	5/5	4/6	2/7	C/T	T/T	
F29	4/9	**4/13**	5/5	**5/5**	**6/9**	C/T	C/T	
P30	**7**	**8**	4	**6**	**10**	C	C	PAT
M30	**7/7**	**8/8**	4/6	**5/6**	**9/10**	C/C	C/C	
F30	9/9	9/9	4/4	5/5	7	C/C	C/C	
P31	7	**8**	4	6	**10**	C	**C**	PAT
M31	7/7	**8/13**	4/5	6/6	**10/10**	C/C	**C/T**	
F31	2/7	13/13	4/5	5/6	2/7	C/T	T/T	
P32	**4**	13	**11**	4	**6**	-	**-**	MAT
M32	5/8	13/13	2/5	3/4	7/7	C/C	C/T	
F32	**4/9**	9/13	**4/11**	4/4	**6/9**	-	C/T	
P33	**8**	**14**	**4**	**6**	**12**	C	T	PAT
M33	**8/11**	**5/14**	**4/4**	**1/6**	**2/12**	C/C	C/T	
F33	2/2	13/13	5/11	5/8	7/7	C/T	T/T	
P34	**3**	**14**	**4**	**4**	**7**	T	C	PAT
M34	**3/3**	**13/14**	**4/5**	**4/4**	**7/7**	C/T	C/T	
F34	2/7	8/13	5/5	5/5	2/2	C/T	C/T	
P35	**5**	**14**	5	6	**7**	C	T	MAT
M35	3/6	10/13	2/5	5/6	6/9	C/T	T/T	
F35	**2/5**	**14/14**	5/10	6/6	**7/7**	C/T	T/T	
P36	**7**	13	**5**	5	**7**	-	-	PAT
M36	**7/7**	9/13	**5/5**	5/5	**7/7**	-	-	
F36	5/5	13/13	7/7	5/6	6/6	-	-	

P37	8	**7**	**5**	5	5	**T**	C	PAT
M37	8/8	**7/16**	**5/5**	5/6	5/10	**C/T**	U	
F37	8/8	8/13	6/8	5/5	5/7	C/C	C/T	
P38	**4**	**13**	-	6	**7**	T	T	MAT
M38	2/7	12/14	6/11	5/6	10/10	C/T	T/T	
F38	**4/4**	**13/13**	4/4	5/6	**2/7**	T/T	T/T	
P39	**4**	**14**	**3**	**6**	7	C	T	PAT
M39	**4/7**	**14/14**	**3/4**	**6/6**	5/7	C/C	T/T	
F39	6/6	6/12	6/11	5/5	7/10	C/T	C/T	
P40	**2**	**13**	4	**5**	**6**	-	-	PAT
M40	**2/2**	**13/13**	4/5	**5/5**	**6/9**	-	-	
F40	-	-	4/4	-	10/11	-	-	
P41	7	**4**	**1**	**6**	8	T	**T**	PAT
M41	4/7	**4/4**	**1/5**	**6/6**	8/11	C/T	**T/T**	
F41	7/10	14/18	6/6	4/4	-	C/T	C/C	
P42	5	**14**	5	**3**	**6**	T	T	MAT
M42	5/7	9/13	5/11	5/5	3/7	T/T	C/T	
F42	2/5	**9/14**	5/5	**3/5**	**6/6**	T/T	C/T	
P43	7	12	**7**	**6**	10	T	T	MAT
M43	4/7	12/12	6/9	1/5	7/10	T/T	T/T	
F43	5/7	11/12	**5/7**	**5/6**	2/10	C/T	T/T	
P44	7	**14**	6	5	-	C	**T**	MAT
M44	4/7	9/17	6/8	5/5	7/10	C/T	C/C	
F44	4/7	**9/14**	6/6	5/6	7/9	C/C	**C/T**	
P45	**2**	**14**	5	**5**	**7**	C	T	PAT
M45	**2/6**	**12/14**	5/5	**1/5**	**7/7**	C/T	-	
F45	4/7	17/17	5/5	6/6	2/10	C/C	C/T	
P46	**7**	**10**	5	**5**	**9**	C	**C**	PAT
M46	**7/7**	**10/13**	4/5	**5/8**	**2/9**	C/T	**C/T**	
F46	4/5	14/14	4/5	6/8	2/10	C/T	T/T	
P47	2	14	**1**	**6**	2	T	T	PAT
M47	2/7	9/14	**1/5**	**6/6**	2/9	C/T	C/T	
F47	2/4	14/14	6/6	4/4	2/2	-	C/T	
P48	**8**	**9**	**4**	6	**11**	T	C	PAT
M48	**5/8**	**6/9**	**4/6**	3/6	**6/11**	T/T	C/C	
F48	7	14/14	2/7	5/6	2/9	C/T	C/T	
P49	**6**	**9**	**6**	**5**	-	**C**	C	MAT
M49	7/7	4/4	5/5	4/4	8/10	T/T	-	
F49	**6/7**	**9/15**	**5/6**	**5/5**	6/7	**C/T**	C/C	
P50	**9**	**6**	**7**	6	**6**	C	C	MAT
M50	**7/7**	**15/17**	**4/5**	6/6	**9/10**	C/T	C/T	
P51	**6**	14	**9**	**6**	-	T	T	MAT
M51	**4/5**	14/17	**5/5**	**5/5**	6/6	T/T	C/T	

P52	7	**15**	4	6	**7**	**T**	T	MAT
M52	7/9	**1/1**	4/5	5/6	**2/9**	**C/C**	C/T	
P53	**7**	**14**	**5**	**5**	**6**	**C**	**T**	PAT
M53	**2/7**	**4/14**	**5/5**	**5/5**	**6/10**	**C/T**	**T/T**	
P54	**7**	**10**	**7**	**6**	9	**C**	**C**	PAT
M54	**7/8**	**10/15**	**6/7**	**6/8**	8	**C/C**	**C/T**	
P55	**7**	15	**5**	**4**	**9**	**C**	**C**	PAT
M55	**7/7**	14/16	**5/5**	**4/4**	**2/9**	**C/T**	**C/T**	
P56	**7**	**9**	**6**	**5**	**8**	-	**T**	PAT
M56	**2/7**	**9/14**	**4/6**	**5/5**	**6/8**	C/T	**T/T**	
P57	**4**	**9**	5	**5**	**6**	C	C	MAT
M57	**5/7**	**12/14**	4/5	**4/6**	**8/10**	C/T	C/C	
P58	**6**	**16**	**6**	**4**	**10**	**T**	C	PAT
M58	**6/8**	**15/16**	**2/6**	**4/5**	**9/10**	**T/T**	-	
P59	7	9	**11**	**9**	10	C	C	MAT
M59	5/7	9/14	**6/7**	**6/6**	6/10	C/T	C/T	
P60	**5**	**14**	**4**	6	**8**	-	-	MAT
M60	**7/12**	**9/9**	**5/6**	3/6	**7/10**	T/T	C/C	
P61	**2**	**4**	**5**	**5**	**7**	**C**	-	PAT
M61	**2/6**	**4/9**	**5/5**	**5/6**	**2/7**	**C/C**	-	

(P) proband, (M) mother, and (F) father; (MAT) Maternal, (PAT) paternal. D22S1638, D22S941, D22S944, D22S1623, D22S264 are microsatellite DNA markers and rs4819519 and rs5993650 are SNP assays. Each allele of the microsatellite markers was numbered according to the fragment size.