

**Additional file 1. Cytogenetic alterations and molecular analysis detailed description**

Patient code	Clinical Manifestations	Karyotype	Molecular Analysis
PM2	Broad BL pattern with disseminated hypo and hyperpigmentation DD, ptosis, scoliosis, low weight and length and DFF	<b>PB:</b> mos 46, XY, r(7) (p22q36.3) [45]/45, XY,-7 [6]/46,XY, dup(7)(p22q36.3)[2] <b>LS:</b> mos 45, XY, -7 [23]/46, XY r(7) (p22q36) [17] <b>DS:</b> mos 45, XY, -7 [26]/46, XY r(7) (p22q36) [29]	<b>1.</b> arr 7p22.3(113,336–954,145)x1,7q36.1q36.3(151306863–158812247) x1, (Human Genome Build, hg18). <b>2.</b> FISH analysis with ToTelVysion DNA probe mixture 7 (Abbot Molecular/Vysis)
PM6	Fine and whorled BL pattern with disseminated hypo and hyperpigmentation DD, seizures and obesity	<b>PB:</b> mos 46, XY, del(18)(q21.3) [3]/46, XY [47] <b>LS:</b> mos 46, XY, del(18)(q21.3) [11]/46, XY [39] <b>DS:</b> mos 46, XY, del(18)(q21.3) [14]/46, XY [36]	NO
PM8	Fine and whorled BL pattern with disseminated hyperpigmentation, DD, hip dysplasia and low weight and length	<b>PB:</b> mos 69,XXY [2]/46,XY [49] <b>LS:</b> 46, XY [50] <b>DS:</b> mos 69,XXY [6]/46,XY [44]	NO
PM9	Fine and whorled BL pattern with disseminated hyperpigmentation, DD, hypotonia, DFF and RHH	<b>PB:</b> mos 46,XX, r(22) (p11.2q13.2)[15]/46,XX [40] <b>LS:</b> mos 46, XX, r(22)(p11.2q13.2) [19]/46, XX [81]	NO
PM13	Fine and whorled BL pattern with disseminated hyperpigmentation, DD and seizures	<b>PB:</b> 46, XY, r(22)(p11.2q13.3) [100] <b>LS:</b> mos 46, XY, r(22)(p11.2q13.3) [98]/46, XY, idic (22) [2] <b>DS:</b> mos 46, XY, r(22)(p11.2q13.3) [96]/46, XY, idic (22) [4]	NO
PM15	Fine and whorled BL pattern with disseminated hyperpigmentation, severe DD, seizures, strabismus and digital alterations	<b>PB:</b> 46, XY [50] <b>LS:</b> mos 47, XY, +i(12)(p10) [11]/46, XY [39] <b>DS:</b> mos 47, XY, +i(12)(p10) [20]/46, XY [30]	NO
PM25	Fine and whorled BL pattern with disseminated hypo and hyperpigmentation, DD, DFF, scoliosis, hip dysplasia and low weight	<b>PB:</b> mos 47,XX,+mar[45]/47,XX, +14[10]/46,XX[45]. <b>LS:</b> mos 47,XX,+mar[7]/46,XX[8] <b>DS:</b> mos 47,XX,+mar[12]/46,XX[14]	<b>1.</b> arr 14q11.1q11.2(18,127,052-19,927,052)x2~3, (UCSC, hg18) <b>2.</b> FISH with DNA BACs probes for 14q11.2 spectrum green and 14q32.33 spectrum <b>Final result:</b> PB: mos 47,XX,+del(14)(q11.2)[45]/47,XX,+14[10]/46,XX[45]
PM27	Fine and whorled BL pattern with disseminated hypo and hyperpigmentation, DFF and digital alterations	<b>PB:</b> 46,XX [50] <b>LS:</b> 46,XX [50] <b>DS:</b> mos 46, XX, t(1;8)(p?36;q?22)[11]/46,XX [139]	NO
PM28	Fine and whorled BL pattern with disseminated hyperpigmentation, DD, seizures, hypotonia, DFF and LHH	<b>PB:</b> mos 47,XY, +mar [4]/46, XY [47] <b>LS:</b> 46, XY [25] <b>DS:</b> 46, XY [50]	<b>1.</b> FISH with $\alpha$ -satellite 15 probe negative (Abbot Molecular/Vysis)

<b>PM30</b>	Fine and whorled BL pattern with disseminated hypo and hyperpigmentation, severe DD, seizures, hypotonia and DFF	<b>PB:</b> mos 47, XX,+mar [28]/45,X [6] / 46, XX [16] <b>LS:</b> mos 45,X [35] / 47, XX,+mar [2]/ 46, XX [6] <b>DS:</b> mos 45,X [20] / 47, XX,+mar [3]/ 46, XX [13]	<b>1.</b> arr Xp22.33q28(60,814-155,254,881)x1[~30%], Xp21.1p11.1(36,025,401-58,483,247)x3 [~55%] (Human Genome Build 37, hg19) <b>2.</b> FISH with $\alpha$ -satellite X probe (AbbotMolecular/Vysis) and locus specific probe (p11.22-p11.23) (Agilent Tech, SureFISH) positive. <b>Final result:</b> PB: mos 47, XX, +der(X)(p21.1p11.1) [28]/45,X [6] / 46, XX [16]
<b>PM39</b>	Fine and whorled BL pattern with disseminated hypo and hyperpigmentation, DD and low height	<b>PB:</b> mos 47,XY,+ mar [44]/ 46,XY [6] <b>LS:</b> mos 47,XY,+ mar [2]/ 46,XY [38] <b>DS:</b> mos 47,XY,+ mar [5]/ 46,XY [45]	<b>1.</b> FISH with $\alpha$ -satellite 14 probe (AbbotMolecular/Vysis) positive <b>2.</b> arr (1-22)x2,(XY)x1, (UCSC, hg19) negative <b>Final result:</b> marker probably of heterochromatic material
<b>PM41</b>	Fine and whorled BL pattern with disseminated hypopigmentation, severe DD, seizures, hypotonia, DFF scoliosis and low weight	<b>PB:</b> 46, X, t(X;22)(q11.2;q13.3) [50] <b>LS:</b> 46, X, t(X;22)(q11.2;q13.3) [50] <b>DS:</b> 46, X, t(X;22)(q11.2;q13.3) [50]	NO
<b>PM42</b>	Fine and whorled BL pattern with disseminated hypopigmentation, DD, seizures, DFF, strabismus, digital alterations and low weight and height,	<b>PB:</b> 46,XY,9qh- [50] <b>LS:</b> mos 47, XY, 9qh-,+mar [19]/ 46,XY, 9qh- [33] <b>DS:</b> mos 47, XY, 9qh-,+mar [9]/ 46,XY, 9qh- [42]	<b>1.</b> arr 9p12p11.2(43,504,857-44,259,464)x1, 9q21.11(69,092,561-69,972,419)x1, (UCSC, hg19) <b>Final result:</b> marker probably of heterochromatic material
<b>PM46</b>	Fine and whorled BL pattern with localized hyperpigmentation, DD, iris heterochromia, DFF and digital alterations	<b>SP:</b> 46,XX, del(13)(q21.3q32.1) [50] <b>LS:</b> 46,XX, del(13)(q21.3q32.1) [50] <b>DS:</b> 46,XX, del(13)(q21.3q32.1) [50]	<b>1.</b> arr 13q21.31q32.1(63,958,459-96,671,274)x1, (UCSC, hg19)
<b>PM52</b>	Fine and whorled BL pattern with disseminated hypo and hyperpigmentation, DD, DFF and low height	<b>PB:</b> 46,XX,22pstk- [50] <b>LS:</b> 46,XX,22pstk- [50] <b>DS:</b> mos 47,XX, +12,22pstk-[44]/46,XX,22pstk- [6]	NO
<b>PM53</b>	Fine and whorled BL pattern with disseminated hyperpigmentation and DD	<b>PB:</b> 46,XX,del(4)(p16.1p15.3) [50] <b>LS:</b> 46,XX,del(4)(p16.1p15.3) [50] <b>DS:</b> 46,XX,del(4)(p16.1p15.3) [50]	<b>1.</b> arr 4p16.1p15.32 (10,047,353-17,614,303)x1, 8p22p21.3 (18,712,712-19, 523, 339)x3, (UCSC, hg19)
<b>PM58</b>	Fine and whorled BL pattern with localized hyperpigmentation, severe DD, hypotonia, DFF and digital alterations	<b>PB:</b> 46,XY [50] <b>LS:</b> mos 47, XY,+12[29]/ 46,XY [21] <b>DS:</b> mos 47, XY,+12[32]/ 46,XY [19]	NO
<b>PM61</b>	Fine and whorled BL pattern with localized hyperpigmentation, DD, microphthalmia, DFF and	<b>PB:</b> 46, XX [50] <b>LS:</b> 46, XX,-18,+mar [50] <b>DS:</b> mos 46, XX,-18,+mar [45]/46, XX [5]	<b>1.</b> FISH FISH with $\alpha$ -satellite 18 probe (AbbotMolecular/Vysis) positive <b>Final result:</b> 46,XX,-18, +mar.ish

	obesity		der(18)(D18Z1+)
PM65	Fine and whorled BL pattern with disseminated hyperpigmentation, DD and low weight	<b>PB:</b> mos 47,XY,+mar [48]/46,XY [2] <b>LS:</b> mos 47,XY,+mar [46]/46,XY [4] <b>DS:</b> mos 47,XY,+mar [27]/46,XY [23]	<b>1.</b> arr 15q11.1q11.2(20,394,272-22,759,378)x3, 15q11.2q13.2(22,759,378-30,509,325)x4 15q13.2q13.3(30,509,325-32,706,883)x3, (UCSC, hg19)
PM67	Fine and whorled BL pattern with localized hyperpigmentation, autistic features	<b>PB:</b> 47,XY, +mar [1] /46,XY [50] <b>LS:</b> mos 46,XY, +mar1 [1]/47,XY,+mar2 [1]/45,XY,-12,+mar2 [1]/48,XY, +mar1, +mar2 [1]/46,XY [46] <b>DS:</b> mos 44,XY,-1,-18,-20,+mar [1]/46,XY [14]	NO
PM69	Fine and whorled BL pattern, hyperpigmentation, DD, seizures, coloboma and low weight	<b>PB:</b> 46,XX [50] <b>LS:</b> 46,XX [35] <b>DS:</b> mos 47,XX,+?22 [3]/46,XX [47]	<b>1.</b> FISH with locus specific probe (22q11.2q11.2) (AbbotMolecular/Vysis) deletion negative
PM71	Fine and whorled BL pattern with localized hyperpigmentation, DD, LHH, cardiac alterations and low weight and length	<b>PB:</b> 46,XY [50] <b>LS:</b> 46,XY, add(16)(p13.3) [25] <b>DS:</b> mos 46,XY, add(16)(p13.3)[9]/46,XY [47]	NO
PM72	Broad BL pattern with disseminated hyperpigmentation, DD and low length	<b>PB:</b> 46,XY [50] 400-500B <b>LS:</b> mos 47,XY,+12 [9]/46,XY [41] <b>DS:</b> mos 47,XY,+12 [17]/46,XY [33]	NO

**BL:** Blaschko Lines; **DD:** Developmental Delay; **DFF:** Dysmorphic Facial Features; **PB:** Peripheral Blood; **LS:** Light Skin (Hypopigmented); **DS:** Dark Skin (Hyperpigmented); **RHH:** Right hemihypertrophy; **LHH:** Left hemihypertrophy.  
**NO:** Molecular analysis not necessary or not realized yet.