**Supplemental Table 1 Prenatal CNVs detected by CMA among the 528 gravidas with positive NIPT results**

| **No.** | **NIPT** | **CNVs (arr[GRCh37])** | **Size of CNVs (kb)** | **Copy number** | **HI/TS region** | **HI/TS gene** | **Inherited** **or de novo** | **ACMG** **Classification** | **Outcomes** | **Concordance** |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| 1 | chr7p22.2 dup | 7p22.2(2966517\_3334799)x3 | 368 | Gain | / | / | NA | VUS | Born | Full concordance |
| 2 | chr5p14.3 del | 5p14.3(19076288\_19448955)x1 | 373 | Loss | / | / | NA | VUS | Born | Full concordance |
| 3 | chr13q12.11 dup | 13q12.11(22618245\_22999366)x3 | 381 | Gain | / | / | NA | VUS | Born | Full concordance |
| 4 | chr3p26.2p26.1 dup | 3p26.2p26.1(3866046\_4273489)x3 | 407 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 5 | chr15q13.3 dup | 15q13.3(31999632\_32428066)x3 | 428 | Gain | / | / | NA | VUS | Born | Full concordance |
| 6 | chr7q11.21 del | 7q11.21(64612880\_65148399)x1 | 536 | Loss | / | / | NA | VUS | Born | Full concordance |
| 7 | chr20q13.13 del | 20q13.13(46470867\_47125819)x1 | 655 | Loss | / | / | NA | VUS | Born | Full concordance |
| 8 | chr5p15.31 dup | 5p15.31(6752757\_7429552)x4 | 677 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 9 | chr7p21.1 dup | 7p21.1(17592111\_18327081)x3 | 735 | Gain | / | / | Inherited from normal father | VUS | Born | Full concordance |
| 10 | chr13q12.11 dup | 13q12.11(21521989\_21972234)x3 | 450 | Gain | / | / | NA | VUS | Born | Full concordance |
| 11 | chr22q11.21 del | 22q11.21(21059669\_21800471)x1 | 741 | Loss | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 12 | chr22q11.21 del | 22q11.21(21059669\_21800471)x1 | 741 | Loss | / | / | *de novo* | VUS | TOP | Full concordance |
| 13 | chr17q11.2 dup | 17q11.2(28749121\_29516669)x3 | 768 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 14 | chr18p11.32 dup | 18p11.32(1244515\_1802917)x3 | 558 | Gain | / | / | NA | VUS | Born (Premature delivery) | Full concordance |
| 15 | chr15q13.3 del | 15q13.3(31967070\_32914239)x1 | 947 | Loss | 15q13.3 recurrent region (D\_CHRNA7 to BP5) (includes CHRNA7 and OTUD7A) | / | NA | P | Born | Full concordance |
| 16 | chr8q24.11 dup | 8q24.11(117990712\_119012676)x3 | 1022 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 17 | chr13q31.1 dup | 13q31.1(81924090\_83110879)x3 | 1187 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 18 | chr2p12 dup | 2p12(78631710\_79851089)x4 | 1219 | Gain | / | / | NA | VUS | Born | Full concordance |
| 19 | chr13q12.12 del | 13q12.12(23533511\_24970361)x1 | 1437 | Loss | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 20 | chr10q24.32q25.1 dup | 10q24.32q25.1(104583879\_106039196)x3 | 1455 | Gain | / | / | NA | VUS | Born | Full concordance |
| 21 | chr13q32.1 dup | 13q32.1(95340470\_96874757)x3 | 1534 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 22 | chr2q12.2q12.3 del | 2q12.2q12.3(106856366\_108527327)x1 | 1670 | Loss | / | / | NA | VUS | Born | Full concordance |
| 23 | chr16p13.12p13.11 dup | 16p13.12p13.11(14770633\_16538596)x3 | 1768 | Gain | / | / | NA | VUS | Born | Full concordance |
| 24 | chr1q21.1q21.2 del | 1q21.1q21.2(146106724\_147933973)x1 | 1827 | Loss | 1q21.1 recurrent region (BP3\_BP4 distal) (includes GJA5) | / | Inherited from normal mother | P | Born | Full concordance |
| 25 | chr8p23.2p23.1 dup | 8p23.2p23.1(4322453\_6204870)x3 | 1882 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 26 | chr22q11.22q11.23 dup | 22q11.22q11.23(22997928\_24995256)x3 | 1997 | Gain |  | / | *de novo* | VUS | Born | Full concordance |
| 27 | chr7q11.21q11.22 dup | 7q11.21q11.22(66785467\_68970684)x4 | 2185 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 28 | chr5p15.31 dup chr14q31.3 dup | 5p15.31(8827019\_9798033)x3 14q31.3(85359235\_87586936)x4 | 9712228 | GainGain | // | // | Inherited from normal motherInherited from normal mother | VUS VUS | Born | Full concordance |
| 29 | chr16q21 dup | 16q21(61996041\_64301745)x3 | 2306 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 30 | chr22q11.21 del | 22q11.21(18648855\_21062134)x1 | 2413 | Loss | 22q11.2 recurrent (DGS/VCFS) region (proximal A\_B) (includes TBX1) | / | *de novo* | P | TOP | Full concordance |
| 31 | chr18q22.1 dup | 18q22.1(64283071\_66769260)x3 | 2486 | Gain | / | / | NA | VUS | NA | Full concordance |
| 32 | chr21q dup | 21q22.3(43941791\_46523623)x3 | 2582 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 33 | chr22q11.21 dup | 22q11.21(18627051\_21283290)x3 | 2656 | Gain | 22q11.2 recurrent (DGS/VCFS) region (proximal A\_B) (includes TBX1) | / | NA | P | TOP | Full concordance |
| 34 | chr12q24.32q24.33 del | 12q24.32q24.33(127391909\_130111679)x1 | 2720 | Loss | / | / | Inherited from normal father | VUS | Born (Premature delivery) | Full concordance |
| 35 | chr14q21.2q21.3 del | 14q21.2q21.3(46210321\_49016299)x1 | 2806 | Loss | / | / | Inherited from normal mother | VUS | Born (hydronephrosis) | Full concordance |
| 36 | chr22q11.21 dup | 22q11.21(18648856\_21461017)x3 | 2812 | Gain | 22q11.2 recurrent (DGS/VCFS) region (proximal A\_B) (includes TBX1) | / | NA | P | TOP | Full concordance |
| 37 | chr16p13.11p12.3 dup | 16p13.11p12.3(15325073\_18157612)x3 | 2833 | Gain | / | / | NA | VUS | Born | Full concordance |
| 38 | chr22q11.21 del | 22q11.21(18631364\_21800471)x1 | 3169 | Loss | 22q11.2 recurrent (DGS/VCFS) region (proximal A\_D) (includes TBX1) | / | NA | P | TOP | Full concordance |
| 39 | chr6p12.3 dup | 6p12.3(46466383\_50480392)x3 | 4014 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 40 | chr2q12.2q13 del | 2q12.2q13(107040325\_111370025)x1 | 4330 | Loss | / | / | Inherited from normal mother | VUS | Neonatal death | Full concordance |
| 41 | chr16q11.2q12.1 dup | 16q11.2q12.1(46503969\_51098261)x3 | 4594 | Gain | / | / | NA | VUS | Born | Full concordance |
| 42 | chr21q21.1 dup | 21q21.1(18490110\_23347274)x3 | 4857 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 43 | chr15q11.2q13.1 dup | 15q11.2q13.1(23632678\_28560664)x3 | 4928 | Gain | 15q11q13 recurrent (PWS/AS) region (BP2\_BP3 Class 2) | / | Inherited from normal mother | P | Born | Full concordance |
| 44 | chr5q22.1q23.1 dup | 5q22.1q23.1(110896866\_116195651)x3 | 5299 | Gain | / | / | NA | VUS | TOP | Full concordance |
| 45 | chr8q23.1q23.3 dup | 8q23.1q23.3(110273153\_115684011)x3 | 5411 | Gain | / | / | NA | VUS | Born | Full concordance |
| 46 | chr11p15.1p14.3 dup | 11p15.1p14.3(20253705\_25684613)x3 | 5431 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 47 | chr11p15.1p14.3 dup | 11p15.1p14.3(20277669\_25713381)x3 | 5436 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 48 | chr10q11.22q11.23 del | 10q11.22q11.23(46293590\_51817663)x1 | 5524 | Loss | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 49 | chr15q26.1q26.3 dup | 15q26.1q26.3(93404310\_98968661)x3 | 5564 | Gain | / | / | NA | VUS | Born | Full concordance |
| 50 | chr16p13.13p12.3 del | 16p13.13p12.3(12548052\_18242713)x1 | 5695 | Loss | 16p13.11 recurrent region (BP2\_BP3) (includes MYH11) | / | NA | P | Born | Full concordance |
| 51 | chr15q11.2q13.1 dup | 15q11.2q13.1(22770421\_28526905)x3 | 5756 | Gain | 15q11q13 recurrent (PWS/AS) region (BP1\_BP3 Class 1) | / | Inherited from normal mother | P | NA | Full concordance |
| 52 | chr12q21.2q21.31 del | 12q21.2q21.31(79313475\_85441579)x1 | 6128 | Loss | / | PPP1R12A | NA | P | Born | Full concordance |
| 53 | chr8q21.13q21.3 del | 8q21.13q21.3(80275606\_87340145)x1 | 7065 | Loss | / | / | NA | VUS | TOP | Full concordance |
| 54 | chr10q22.3q23.2 dup | 10q22.3q23.2(81674867\_88957815)x3 | 7283 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 55 | chr10q22.3q23.2 del | 10q22.3q23.2(81630469\_88957815)x1 | 7327 | Loss | 10q22.3q23.2 recurrent region (LCR\_3/4\_flanked) (includes BMPR1A) | BMPR1A/PTEN | NA | P | TOP | Full concordance |
| 56 | chr10q22.3q23.2 dup | 10q22.3q23.2(81630469\_88973570)x3 | 7343 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 57 | chr7p21.3p21.1 del | 7p21.3p21.1(8974812\_17191226)x1 | 8216 | Loss | / | / | *de novo* | VUS | TOP | Full concordance |
| 58 | chr11q24.2q25 del | 11q24.2q25(126647772\_134937416)x1 | 8290 | Loss | / | / | *de novo* | VUS | TOP | Full concordance |
| 59 | chr3q11.1q12.3 dup | 3q11.1q12.3(93519465\_101839691)x3 | 8320 | Gain | / | / | Inherited from normal mother | VUS | NA | Full concordance |
| 60 | chr8p23.3p23.1 del | 8p23.3p23.1(158048\_9010029)x1 | 8852 | Loss | / | / | NA | LP | TOP | Full concordance |
| 61 | chr5q21.1q22.1 dup | 5q21.1q22.1(101522649\_110634622)x3 | 9112 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 62 | chr5p11q11.2 dup | 5p11q11.2(46365514\_55986750)x3 | 9621 | Gain | / | / | Inherited from normal mother | VUS | Born | Full concordance |
| 63 | chr2p23.1p22.1 del | 2p23.1p22.1(30733152\_41085497)x1 | 10352 | Loss | / | SPAST | NA | P | Born | Full concordance |
| 64 | chr8p23.3p23.1 del | 8p23.3p23.1(158049\_10685851)x1 | 10528 | Loss | / | / | NA | LP | TOP | Full concordance |
| 65 | chr3q27.2q29 dup | 3q27.2q29(185409997\_197851444)x3 | 12441 | Gain | / | / | NA | LP | TOP | Full concordance |
| 66 | chr5q21.3q23.1 del | 5q21.3q23.1(104832795\_118713574)x1 | 13881 | Loss | / | APC | NA | P | TOP | Full concordance |
| 67 | chr21q11.2q21.3 del | 21q11.2q21.3(15016487\_29188153)x1 | 14172 | Loss | / | / | *de novo* | VUS | NA | Full concordance |
| 68 | chr4q13.1q21.1 del | 4q13.1q21.1(62323426\_76675789)x1 | 14352 | Loss | / | / | *de novo* | P | TOP | Full concordance |
| 69 | chr18q22.1q23 del | 18q22.1q23(63578361\_78013728)x1 | 14435 | Loss | / | EYA1 | Inherited from mother with intellectual disability | P | NA | Full concordance |
| 70 | chr18p11.32 delchr18p11.32p11.21 dup | 18p11.32(136228\_1331930)x1 18p11.32p11.21(1343954\_15170636)x3 | 119613827 | LossGain | // | // | NA | VUS LP | Born | Full concordance |
| 71 | chr18p11.32q23 dup | 18p11.32q23(136228\_15181207).15 | 15045 | Gain (mosaic) | / | / | NA | LP | TOP | Full concordance |
| 72 | chr2q23.3q31.1 dup | 2q23.3q31.1(154358188\_170720261)x3 | 16362 | Gain | / | / | NA | LP | TOP | Full concordance |
| 73 | chr5p15.33p14.3 del | 5p15.33p14.3(113576\_18727376)x1 | 18614 | Loss | 5p15 terminal (Cri du chat syndrome) region | TRIO | NA | P | TOP | Full concordance |
| 74 | chr4q31.3q34.2 del | 4q31.3q34.2(154851387\_176868942)x1 | 22018 | Loss | / | / | NA | P | TOP | Full concordance |
| 75 | chr5p15.33p14.1 del | 5p15.33p14.1(113577\_26243789)x1 | 26130 | Loss | 5p15 terminal (Cri du chat syndrome) region | TRIO | NA | P | TOP | Full concordance |
| 76 | chr18p11.22 dup | 18p11.22(8718383\_10139732)x3 22q11.23q12.1(25116001\_26437690)x3 | 14211322 | GainGain | // | // | Inherited from normal mother*de novo* | VUS VUS | TOP (hydrocephalus) | Partial concordance |
| 77 | chr8p23.3p23.2 del | 8p23.3p23.2(158048\_3220759)x1 8q21.11q24.3(77115706\_146295771)x3 | 3063 69180 | LossGain | // | // | Paternal invertion: 46,XY,inv(8)(p23.2;q21) **\*** | VUS LP | TOP | Partial concordance |
| 78 | chr8p23.3p23.2 del | 8p23.3p23.2(158048\_4745371)x1 9p24.3p21.3(208454\_22492876)x3 | 4587 22284 | LossGain | // | // | Maternal balancedtranslocation: 46,XX,t(8;9)(p23;p21.3) | VUS LP | TOP | Partial concordance |
| 79 | chr20p13p12.1 dup | 20p13p12.1(61662\_13546848)x39p24.3p23(208455\_11144684)x1 | 1342610936 | GainLoss | // | // | Paternal balancedtranslocation: 46,XY,t(9;20)(p23;p12.1) | LP LP | TOP | Partial concordance |
| 80 | chr9p24.3p22.3 dup | 9p24.3p22.3(208455\_15608372)x35p15.33(113577\_1641914)x1 | 154001528 | GainLoss | // | // | Inherited from mother with intellectual disability: 46,XX,der t(5;9)(p15.3;p22) | VUS VUS | TOP | Partial concordance |
| 81 | chr12q24.21q24.33 dup | 12q24.21q24.33(116785882\_133777562)x32q37.3(240495629\_242782258)x1 | 169922287 | GainLoss | //  | // | Maternal balancedtranslocation: 46,XX,t(2;12)(q37;q24) | LP VUS | TOP | Partial concordance |
| 82 | chr18q21.33q23 dup | 18q21.33q23(60906988\_78013728)x34q35.2(187316147\_190957460)x1 | 171073641 | GainLoss | //  | // | Inherited from mother with intellectual disability: 46,XX,der t(4;18)(q35;q22) | VUS VUS | TOP | Partial concordance |
| 83 | chr17q23.3q25.3 dup  | 17q23.3q25.3(61775472\_81041823)x3 1p36.33p36.32(849466\_3777765)x1 | 19266 2928 | Gain Loss | / 1p36 terminal region (includes GABRD) | // | Maternal balancedtranslocation: 46,XX,t(1;17)(p36;q23) | LP P | TOP | Partial concordance |
| 84 | chr18q21.32q23 del | 18q21.32q23(58106726\_78013728)x1 1q43(238949246\_240062389)x1 | 19907 1113 | LossLoss | //  | // | NA | LP VUS | TOP | Partial concordance |
| 85 | chr9p24.3p13.2 dup | 9p24.3p13.2(1581286\_37055141)x3 9p24.3(208454\_1577575)x1 | 35474 1369 | GainLoss | //  | // | NA | LP VUS | TOP | Partial concordance |
| 86 | chr20p13p11.21 dup | 20p13p11.21(61661\_24487341)x311q24.2q25(126392021\_134937416)x1 | 244268545 | GainLoss | //  | // | Maternal balancedtranslocation: 46,XX,t(11;20)(q24.2;p11.2) | LP VUS | TOP | Partial concordance |
| 87 | chr3p26.3p22.2 dup | 3p26.3p22.2(285857\_37597219)x3 5p15.33p15.31(113577\_8750244)x1 | 37311 8637 | GainLoss | / 5p15 terminal (Cri du chat syndrome) region | /TRIO | Paternal balancedtranslocation: 46,XY,t(3;5)(p24;p15.3) | VUS P | TOP | Partial concordance |
| 88 | chr14q del | Yq11.21q11.221(14460771\_15220682)x0 | 760 | Loss | / | / | NA | VUS | Born | Discordance |
| 89 | chr7q del | 16p11.2(29428531\_30190029)x1 | 761 | Loss | 16p11.2 recurrent region (proximal BP4\_BP5) (includes TBX6) | / | *de novo* | P | TOP | Discordance |
| 90 | chr1p del chr4q dup | 4q35.2(190114475\_190957460)x1 | 843 | Loss | / | / | NA | VUS | Born | Discordance |
| 91 | chr15q dup | 10p13p12.33(17066844\_18286639)x3 | 1220 | Gain | / | / | Inherited from normal mother | VUS | Born | Discordance |
| 92 | chr7q11.22 del | Xp22.31(6449837\_8143509)x1 | 1694 | Loss | Xp22.31 recurrent region (includes STS) | STS | NA | P | Born | Discordance |
| 93 | chr18p11.23 dup | 4q34.3(178130291\_179860825)x3 | 1731 | Gain | / | / | NA | VUS | Born | Discordance |
| 94 | chr5q15 dup | 8q24.12(119740530\_122337637)x3 | 2597 | Gain | / | / | Inherited from normal father | VUS | Born | Discordance |
| 95 | chr11p13 del | 1q25.3q31.1(184825946\_187563410)x1 | 2737 | Loss | / | / | Inherited from normal mother | VUS | Born | Discordance |
| 96 | chr14q24.1 dup | 5p15.1p14.3(17897900\_21148212)x3 | 3250 | Gain | / | / | Inherited from normal mother | VUS | Born | Discordance |
| 97 | chr14q24.3 del | 5p15.33p15.32(113577\_6138632)x1 | 6025 | Loss | 5p15 terminal (Cri du chat syndrome) region | TRIO | NA | P | TOP | Discordance |

CNV: copy number variation; CMA: chromosomal microarray analysis; NIPT: noninvasive prenatal testing; HI: haploinsufficiency; TS: triplosensitivity; ACMG: American College of Medical Genetics and Genomics; NA: not available; P: pathogenic; LP: likely pathogenic; VUS: uncertain clinical significance; TOP: termination of pregnancy

**Supplemental Table 2 Prenatal ROHs detected by CMA among the 528 gravidas with positive NIPT results**

| **No.** | **NIPT** | **CNVs (arr[GRCh37])** | **Size of ROHs (kb)** | **Copy number** | **Disorder** | **Source** | **Outcomes** |
| --- | --- | --- | --- | --- | --- | --- | --- |
| 98 | trisomy 1 | 1p36.33p11.2(888658\_121339317)x2 hmz1q21.2q44(149879544\_249198164)x2 hmz | 12045199319 | UPD1 (isodisomy) | / | NA | TOP |
| 99 | trisomy 1 | 1p36.33p11.2(888658\_121339317)x2 hmz1q21.2q44(149879544\_249198164)x2 hmz | 12045199319 | UPD1 (isodisomy) | / | NA | TOP (FGR) |
| 100 | trisomy 1 | 1p36.33p11.2(888658\_121339317)x2 hmz1q21.2q44(149879544\_249198164)x2 hmz | 12045199319 | UPD1 (isodisomy) | / | NA | Born (Methylmalonic acidemia) |
| 101 | trisomy 2 | 2p25.3p11.2(50813\_87053152)x2 hmz 2q11.1q37.3(95550957\_242773583)x2 hmz | 87002147223 | UPD2 (isodisomy) | / | NA | TOP (FGR) |
| 102 | trisomy 2 | 2p25.3p11.2(50813\_87053152)x2 hmz2q11.1q37.3(95550957\_242773583)x2 hmz | 87002147223 | mUPD2 (isodisomy) | / | maternal  | TOP (hydrops fetalis) |
| 103 | trisomy 2 | 2p24.1p16.1(19693805\_59685825)x2 hmz2q14.2q24.1(121278207\_158756848)x2 hmz2q31.1q34(175042562\_213345197)x2 hmz | 399923747938303 | mUPD2 (iso\_heterodisomy\*) | / | maternal | Born |
| 104 | trisomy 3 | 3p26.3p25.1(73602\_16294894)x2 hmz3p12.3p11.1(74601403\_88586090)x2 hmz3q11.1q13.11(93558925\_105429152)x2 hmz | 162211398511870 | mUPD3 (iso\_heterodisomy) | / | maternal | NA |
| 105 | trisomy 4 | 4p16.3p11(75173\_49063479)x2 hmz 4q11q35.2(52696791\_190921709)x2 hmz | 48988138225 | UPD4 (isodisomy) | / | NA | TOP |
| 106 | trisomy 4 | 4p16.3p11(75173\_49063479)x2 hmz4q11q35.2(52696791\_190921709)x2 hmz | 48988138225 | pUPD4 (isodisomy) | / | paternal | TOP (FGR) |
| 107 | trisomy 4 | 4p16.3p11(75173\_49063479)x2 hmz4q11q35.2(52696791\_190921709)x2 hmz | 48988138225 | UPD4 (isodisomy) | / | NA | Born |
| 108 | trisomy 4 | 4p16.3p15.33(75173\_15121280)x2 hmz4p14p11(38705256\_49063479)x2 hmz4q28.2q34.2(129685157\_177412472)x2 hmz | 150461035847727 | mUPD4 (iso\_heterodisomy) | / | maternal | TOP |
| 109 | trisomy 6 | 6p25.3p11.1(203877\_58726706)x2 hmz6q11.1q27(61972917\_170896644)x2 hmz | 58523 108924 | pUPD6 (isodisomy) | Transient NeonatalDiabetes mellitus (TNDM) | paternal | TOP (FGR) |
| 110 | trisomy 6 | 6p25.3p11.1(203877\_58726706)x2 hmz6q11.1q27(61972917\_170896644)x2 hmz | 58523 108924 | mUPD6 (isodisomy) | / | maternal | Born |
| 111 | trisomy 6 | 6p25.3p11.1(203877\_58726706)x2 hmz6q11.1q27(61972917\_170896644)x2 hmz | 58523 108924 | UPD6 (isodisomy) | / | NA | TOP |
| 112 | trisomy 6 | 6p24.1p11.1(13395610\_58726706)x2 hmz6q16.1q22.31(94446431\_124307093)x2 hmz6q25.1q27(150710446\_170896644)x2 hmz | 453312986120186 | mUPD6 (iso\_heterodisomy) | / | maternal | TOP (FGR) |
| 113 | trisomy 6 | 6p25.3p11.1(203878\_58726706)x2 hmz 6q11.1q27(61972918\_170896644)x2 hmz | 58523 108924 | mUPD6 (isodisomy) | / | maternal | Born |
| 114 | trisomy 7 | 7p22.3p11.1(50943\_58019983)x2 hmz 7q11.21q36.3(62569501\_159118443)x2 hmz | 5796996549 | UPD7 (isodisomy) | / | NA | Born (short stature) |
| 115 | trisomy 7 | 7p22.3p11.1(50943\_58019983)x2 hmz7q11.21q36.3(62569501\_159118443)x2 hmz | 5796996549 | UPD7 (isodisomy) | / | NA | TOP |
| 116 | trisomy 8 | 8p23.1p11.1(8117564\_43776564)x2 hmz8q11.1q24.3(46919156\_146292734)x2 hmz | 3565999374 | mUPD8 (isodisomy) | / | maternal | TOP |
| 117 | trisomy 8 | 8p23.1p11.1(8117564\_43776564)x2 hmz8q11.1q24.3(46919156\_146292734)x2 hmz | 3565999374 | UPD8 (isodisomy) | / | NA | TOP |
| 118 | trisomy 9 | 9p24.3p13.1(216123\_38771831)x2 hmz 9q21.11q34.3(71013799\_141011581)x2 hmz | 38556 69998 | UPD9 (isodisomy) | / | NA | TOP |
| 119 | trisomy 9 | 9p24.3p13.1(216124\_38771831)x2 hmz9q21.11q34.3(71013800\_141011581)x2 hmz | 3855669998 | pUPD9 (iso\_heterodisomy) | / | paternal | TOP |
| 120 | trisomy 11 | 11q23.2q25(112793095\_134930689)x2 hmz | 22138 | mUPD11 (iso\_heterodisomy) | Silver–Russellsyndrome (SRS) | maternal | TOP |
| 121 | trisomy 11 | 11q14.2q25(88151706\_134930689)x2 hmz | 46779 | pUPD11 (iso\_heterodisomy) | Beckwith–Wiedemannsyndrome (BWS) | paternal | TOP |
| 122 | chr14q23.3q24.3 dup dup | 14q23.1q32.11(61630461\_91714413)x2 hmz | 30084 | mUPD14 (iso\_heterodisomy) | Temple syndrome | maternal | TOP |
| 123 | chr15q21.2 dup | 15q21.1q22.2(48078591\_62709924)x2 hmz | 14631 | mUPD15 (iso\_heterodisomy) | Prader–Willisyndrome (PWS) | maternal | TOPPlacental trisomy 15 confirmed by FISH |
| 124 | trisomy 16 | 16p13.3p13.13(94808\_11870494)x2 hmz 16q23.1q24.3(77786018\_90146366)x2 hmz | 1177612360 | mUPD16 (iso\_heterodisomy) | / | maternal | TOP |
| 125 | trisomy 16 | 16p13.3p12.3(94807\_20050658)x2 hmz 16q22.1q24.3(69860932\_90146366)x2 hmz | 1995620285 | mUPD16 (iso\_heterodisomy) | / | maternal | Born |
| 126 | trisomy 16 | 16p13.3p13.13(94807\_11219041)x2 hmz16q22.3q24.3(73469057\_90146366)x2 hmz | 1112411667 | mUPD16 (iso\_heterodisomy) | / | maternal | TOP (demise)Placental trisomy 16 confirmed by FISH |
| 127 | trisomy 17 | 17p13.3p11.2(18900\_22170994)x2 hmz17q11.1q25.3(25309336\_81041760)x2 hmz | 2215255732 | UPD17 (isodisomy) | / | NA | TOP (FGR) |
| 128 | trisomy 1 | 1p13.3p11.2(108503808\_121339317)x2 hmz1q21.2q24.3(149879544\_172597553)x2 hmz1q41q43(218237293\_241346599)x2 hmz | 128362271823109 | ROH | / | NA | Born |
| 129 | trisomy 1 | 1p32.2p21.3(58259167\_99399687)x2 hmz1q25.3q42.12(182520611\_226528744)x2 hmz | 4114144008 | ROH | / | NA | NA |
| 130 | trisomy 1 | 1p13.2p11.2(115835500\_121339317)x2 hmz1q21.2q23.3(149879545\_165480347)x2 hmz | 550415601 | ROH | / | NA | Born |
| 131 | trisomy 2 | 2p25.3p22.2(1989288\_37998488)x2 hmz 2q36.1q37.3(223571444\_242773583)x2 hmz | 36009 19202 | ROH | / | NA | Born (short stature) |
| 132 | trisomy 2 | 2p13.2p11.2(72382277\_87053152)x2 hmz2q11.1q12.3(95550957\_109616111)x2 hmz | 1467114065 | ROH | / | NA | Neonatal death |
| 133 | trisomy 2 | 2p12p11.2(75698714\_87053152)x2 hmz2q11.1q12.2(95550957\_106174659)x2 hmz | 1135410624 | ROH | / | NA | Born |
| 134 | trisomy 3 | 3p26.3p26.1(73603\_6891874)x2 hmz 3p22.2p13(39251141\_69830674)x2 hmz 3q25.1q27.2(150678233\_185795060)x2 hmz | 6818 3058035117 | ROH | / | NA | TOP |
| 135 | trisomy 6 | 6p25.3p11.1(203877\_58726706)x2 hmz | 58523 | ROH | / | NA | TOP (LVOTO) |
| 136 | trisomy 8 | 8p12p11.1(31079982\_43776564)x2 hmz8q11.1q12.2(46919156\_61854841)x2 hmz | 1269714936 | ROH | / | NA | Born |
| 137 | trisomy 8 | 8p21.3p11.23(20832793\_36638339)x2 hmz8q12.3q22.1(65927736\_93643205)x2 hmz | 1580627715 | ROH | / | NA | NA |
| 138 | trisomy 8 | 8p12p11.1(34070872\_43776564)x2 hmz8q11.1q13.2(46919157\_68771501)x2 hmz | 9705721852 | ROH | / | NA | Born |
| 139 | trisomy 9 | 9p24.3p21.3(216123\_20570700)x2 hmz9q31.3q34.3(112814078\_141011581)x2 hmz | 2035528198 | ROH | / | NA | TOP |
| 140 | trisomy 12 | 12p13.33p11.22(257935\_30396571)x2 hmz | 30139 | ROH | / | NA | Born |
| 141 | trisomy 13 | 13q31.1q34(84417821\_115095705)x2 hmz | 30678 | ROH | / | NA | Born |
| 142 | trisomy 16 | 16p13.3p13.13(94808\_12292798)x2 hmz 16q23.2q24.3(80394565\_90146366)x2 hmz | 12198 9752 | ROH | / | NA | NA |
| 143 | trisomy 16 | 16q23.1q24.3(78950254\_90146366)x2 hmz | 11196 | ROH | / | NA | Born |
| 144 | trisomy 16 | 16p13.3p13.12(94808\_14053831)x2 hmz | 13959 | ROH | / | NA | TOP |
| 145 | trisomy 16 | 16p13.3p12.3(94808\_19331243)x2 hmz | 19236 | ROH | / | NA | Born |
| 146 | trisomy 16 | 16p13.3(94808\_7154181)x2 hmz 16q23.3q24.3(81940867\_90146366)x2 hmz | 70598206 | ROH | / | NA | Born |
| 147 | trisomy 16 | 16p13.3(94808\_5500174)x2 hmz 16q21q23.3(66159040\_83117017)x2 hmz | 540516958 | ROH | / | NA | fetal loss after amniocentesis |

CMA: chromosomal microarray analysis; NIPT: noninvasive prenatal testing; UPD:uniparental disomy; ROH: regions of homozygosity; NA: not available; TOP: termination of pregnancy; FGR: fetal growth restriction; LVOTO: left ventricular outflow tract; FISH: fluorescence in situ hybridization

\*: Iso\_heterodisomy indicates combined isodisomy and heterodisomy (mixtures of both subtypes).