

“A retrospective single center analysis of fetuses with region of homozygosity detected by single nucleotide polymorphism array”. The paper was coauthored by Xiufen Bu, Xiuyun Yu, Li Zeng,Guo Zeng, Rong Tan, Can Peng, Shihao Zhou, Siyuan Linpeng and Jing Liu.

Supplementary Table S1. Fifty-five fetuses with ROH on nonimprinted chromosomes but on copy number alterations.

NO.	Case number	Maternal Age	GA	Indication	ROH (GRCh37) detected by SNP array	Size (Mb)	Results of further genetic testing	Pregnancy outcome
1	Case 1	36 y	22 w+	NIPT indicated a 69.3 Mb duplication at 2p11.2p25.3, FGR, advanced maternal age	2q31.1q37.3(174605494_242773583)x2 hmz	68.1	Normal karyotype	TOP, FGR, oligohydramnios, increased placental thickness
2	Case 2	31 y	18 w+	High risk of T16 detected by NIPT	16p13.3p12.3(94807_17705580)x2 hmz,16q22.3q24.3(73772289_90146366)x2 hmz	17.6 16.3	Normal karyotype	Term birth, BW 2.85 kg, healthy
3	Case 4	32 y	21 w+	High risk of T13 detected by NIPT	11q12.1q13.3(56541387_68592901)x2 hmz	12.0	Normal karyotype	Term birth, BW 2.8 kg, healthy
4	Case 8	38 y	18 w+	NIPT indicated a 17.3 Mb deletion at 5p15.1p15.33	5p15.33p15.1(113576_16203210)x2 hmz	16.0	Normal karyotype	Term birth, BW 2.6 kg, healthy
5	Case 12	38 y	20 w+	Advanced maternal age	12q21.2q21.32(78575542_88808147)x2 hmz	10.2	Normal karyotype	Term birth, BW 3.6 kg, healthy
6	Case 15	40 y	19 w+	Advanced maternal age	12q15q21.33(69961050_90562017)x2 hmz	20.6	Normal karyotype	Term birth, BW 3.65 kg, healthy
7	Case 16	29 y	16 w+	NIPT indicated a 20.17 Mb duplication at 8p23.2p21.2	8p23.3p11.1(168483_43776564)x2 hmz	43.6	parental SNP-array indicated maternal isoUPD8p	Term birth, BW 2.15 kg, healthy
8	Case 17	41 y	16 w+	Advanced maternal age	2q21.3q22.3(136750789_147085498)x2 hmz	10.3	46,XYqh- (Y=22)	Term birth, BW 3kg, healthy
9	Case18	46 y	18 w+	Advanced maternal age, high risk of maternal serum screening	3p26.3q29(73602_197791601)x2 hmz	194	parental SNP-array indicated paternal isoUPD3	Term birth, BW 2.6 kg, healthy
10	Case 19	30 y	18 w+	High risk of maternal serum screening	2q22.1q24.1(140922099_158925959)x2 hmz, 12q13.13q14.2(52965781_64150146)x2 hmz	18.0 11.2	Normal karyotype	Term birth, BW 4 kg, healthy
11	Case 20	21 y	18 w+	High risk of maternal serum screening	2q24.3q32.2(165349901_190443583)x2 hmz	25.1	Normal karyotype	Term birth, BW 3.6 kg
12	Case 21	24 y	23 w+	Strephenopodia	5q15q21.2(92745387_103542531)x2 hmz	10.8	Normal karyotype	Term birth, BW 2.7 kg
13	Case 22	27 y	30 w+	Small FL and HL for gestation age, ventricular septal defect, subependymal cyst	19q13.11q13.31(34144743_44168281)x2 hmz	10.0	/	Term birth, BW 2.8 kg, healthy
14	Case 23	30 y	18 w+	NIPT indicated a 10.7 Mb deletion at 1p36.33p36.22,	1p36.33p36.22(888658-10454773)x2 hmz	9.57	Normal karyotype	Term birth, BW 2.6 kg, healthy

				adverse pregnancy history				
15	Case 24	36 y	22 w+	Advanced maternal age, high risk of T16 detected by NIPT	16q23.1q24.3(74839519_90146366)x2 hmz	15.31	Normal karyotype	Term birth, healthy
16	Case 25	37 y	33 w+	Advanced maternal age, high risk of maternal serum screening, increased echogenicity of fetal bowel	12q24.21q24.32(114933859_127616101)x2 hmz	12.68	Normal karyotype	Term birth, healthy
17	Case 26	24 y	17 w+	Abdominal cystic mass, right choroid plexus cyst	2q33.3q35(207785911_218026210)x2 hmz	10.24	Normal karyotype	Term birth, BW 3.2 kg, healthy
18	Case 27	38 y	18 w+	Advanced maternal age, adverse pregnancy history	9q22.2q32(91920199_116462317)x2 hmz	24.54	Normal karyotype	Term birth, BW 2.5 kg, healthy
19	Case 28	38 y	20 w+	High risk of sex chromosome aneuploidy detected by NIPT	18q11.2q21.2(35556032_48297450)x2 hmz	12.74	Normal karyotype	Term birth, BW 1.9 kg, healthy
20	Case 29	18 y	19 w+	high risk of maternal serum screening	18q11.1q12.1(18552516_28949901)x2 hmz	10.4	Normal karyotype	Term birth, BW 2.65 kg, healthy
21	Case 30	35 y	18 w+	Increased echogenicity of fetal bowel, high risk of maternal serum screening	3p25.1p24.1(16053135_27940253)x2 hmz	11.89	46,XY,1qh+	Term birth, BW 3.5 kg, healthy
22	Case 31	29 y	22 w+	NIPT indicated a deletion at 22q11.2	1p31.3p31.1(61906515_81603516)x2 hmz	19.7	Normal karyotype	Term birth, BW 2.65 kg, healthy
23	Case 32	32 y	17 w+	High risk of T2 detected by NIPT, high risk of maternal serum screening	2q21.2q31.1(134110923_176890967)x2 hmz	42.78	Normal karyotype	Term birth, BW 3 kg, healthy
24	Case 34	26 y	18 w+	NIPT indicated a 15.56 Mb deletion at 18q21.32q23	18q21.32q23(56947979_77997606)x2 hmz	21.05	Normal karyotype	Term birth, BW 2 kg, healthy
25	Case 35	31 y	27 w+	Small for gestation age, increased echogenicity of fetal bowel, tricuspid regurgitation	2q32.2q35(189505468_215832746)x2 hmz	26.33	Normal karyotype	Term birth, BW 2.7 kg, healthy
26	Case 36	37 y	23 w+	Decreased amniotic fluid, advanced maternal age	4q28.1q32.1(128181461_159638966)x2 hmz	31.46	Normal karyotype	Term birth, BW 3.1 kg, healthy
27	Case 37	43 y	18 w+	Advanced maternal age, adverse pregnancy history	1p22.3p21.2(86905189_101810361)x2 hmz, 13q21.33q34(71305075_113582441)x2 hmz, 22q12.1q13.31(28794068_44703502)x2 hmz	14.91 42.28 15.91	Normal karyotype	Term birth, BW 2.9 kg, healthy
28	Case 39	30 y	19 w+	NIPT indicated a 40.4 Mb deletion at 8p23.3p11.21	8p23.3p11.1(168483_43776564)x2 hmz	43.6	46,XY,t(6;8)(q25;q22)	Term birth, BW 3 kg, healthy
29	Case 40	29 y	19 w+	High risk of maternal serum screening	1p31.1p13.3(78192652_107501286)x2 hmz	29.31	Normal karyotype	Term birth, BW 3.1 kg, healthy
30	Case 44	25 y	19 w+	High risk of T13 detected by NIPT	13q11q13.3(19450956_35535238)x2 hmz	16.08	Normal karyotype	Term birth, BW 3.25 kg, healthy
31	Case 45	27 y	17 w+	High risk of T16 detected by NIPT	16q22.1q24.3(69837155_90146366)x2 hmz	20.31	Normal karyotype	Preterm birth, FGR, early death

32	Case 47	31 y	18 w+	High risk of maternal serum screening	16p13.3p12.3(94807_17123847)x2 hmz	17.03	Normal karyotype	Term birth, BW 2.5 kg, healthy
33	Case 48	38 y	24 w+	High risk of T16 detected by NIPT, FGR	16p13.3p12.2(94808_23024688)x2 hmz	22.9	parental SNP-array indicated maternal mixUPD16, single-WES indicated no clinically relevant mutations	Preterm birth, BW 1.35 kg, catch-up growth at 2 y, healthy
34	Case 49/Case 50	35 y	22 w+	Monochorionic twin pregnancy, small AC and FL for gestation age	3p25.2p21.1(13267771_53741287)x2 hmz	40.47	parental SNP-array indicated IBD	TOP
35	Case 51	30 y	18 w+	NIPT indicated a 11.22 Mb deletion at 18p11.32p11.21	18p11.32p11.21(136305_11807701)x2 hmz	11.67	Normal karyotype	Term birth, BW 3.2 kg, healthy
36	Case 52	30 y	33 w+	Small HL and FL for gestation age	1q25.3q32.1(184338545_201236141)x2 hmz, 5p14.3p13.3(19788744_30152784)x2 hmz	16.90 10.36	/	Term birth, BW 2.6 kg, healthy
37	Case 53	29 y	18 w+	High risk of sex chromosome aneuploidy detected by NIPT	2p16.3p14(52826571_65727567)x2 hmz	12.9	Normal karyotype	Term birth, BW 3.6 kg, healthy
38	Case 54	31 y	17 w+	NIPT indicated a 18.1Mb deletion at 1p36.33p36.13, high risk of maternal serum screening	1p36.33p36.13(888659_19500139)x2 hmz	18.61	Normal karyotype	Unexplained premature rupture of membranes
39	Case 56	38 y	16 w+	Advanced maternal age, high risk of maternal serum screening	1p36.33q44(849467_249224684)x2 mos hmz	248	Normal karyotype	Term birth, BW 3.5 kg, healthy
40	Case 58	25 y	18 w+	Fetal choroid plexus cysts, high risk of sex chromosome aneuploidy detected by NIPT	8q22.1q24.13(98390954_123676863)x2 hmz	25.29	Normal karyotype	Term birth, BW 2.8 kg, healthy
41	Case 59	21 y	24 w+	Oligohydramnios, ventricular septal defect, coarctation of aorta	2q32.3q35(196375520_218062296)x2 hmz	21.69	Normal karyotype	Miscarriage after amniotic fluid infusion
42	Case 60	31 y	21 w+	Adverse pregnancy history	2p25.3p24.2(50814_16806775)x2 hmz, 4q21.23q32.3(84132874_169686795)x2 hmz, 8p21.3q13.3(20271413_70503919)x2 hmz, 19q12q13.43(32154657_58955556)x2 hmz	16.76 85.56 26.8	Normal karyotype, <i>F</i> value=0.815, trio-WES indicated no clinically relevant mutations	Term birth, BW 3.9 kg, healthy
43	Case 65	28 y	17 w+	Increased nuchal translucency (3.8 mm)	16q11.2q13(46504467_57199560)x2 hmz	10.7	Normal karyotype	Term birth, BW 3kg, healthy
44	Case 67	33 y	18 w+	NIPT indicated a 19.69 Mb deletion at 18q21.31q23	18q21.31q23(54488599_77997606)x2 hmz	23.51	Normal karyotype	Term birth, BW 3.25 kg, healthy
45	Case 71	32 y	19 w+	High risk of maternal serum screening	3p23p21.31(31875826_46656351)x2 hmz	14.78	Normal karyotype	Term birth, BW 3.15 kg, healthy

				Advanced maternal age, high risk of maternal serum screening	1q32.3q42.12(214338164_225575588)x2 hmz, 5p14.3p11(20835592_46313469)x2 hmz, 5q22.1q23.2(110312510_121640434)x2 hmz, 10q21.1q21.3(55295059_65576676)x2 hmz	11.24 25.48 11.33 10.28	Normal karyotype, <i>F</i> -value =0.302	
46	Case 75	38 y	20 w+					Lost to follow-up
47	Case 76	28 y	24 w+	Right hand polydactyly of fetus	1p33p31.3(48288541_62819617)x2 hmz	14.54	Normal karyotype, trio-WES indicated no clinically relevant mutations	Term birth, BW 3.2 kg, healthy
48	Case 77	34 y	18 w+	High risk of T21 detected by NIPT	2q22.3q24.3(148408694_167977820)x2 hmz	19.57	Normal karyotype	Term birth, BW 3.45 kg, healthy
49	Case 80	25 y	24 w+	FGR, VSD, single umbilical artery	16p13.3p13.13(94808_12307880)x2 hmz	12.2	parental SNP-array indicated maternal mixUPD16	Term birth, BW 2.4 kg, healthy
50	Case 81	33 y	17 w+	High risk of T8 detected by NIPT, high risk of maternal serum screening	8p21.2q22.1(26907907_96521988)x2 hmz	19.57	Normal karyotype, single-WES indicated a heterozygous mutation, NM_004260.3: c.2752G>T (P), in <i>RECQL 4</i> , AR	TOP, intrauterine distress, hypercarbonic anemia
51	Case 82	34 y	17 w+	Adverse pregnancy history	2q12.1q23.3(102950822_154399249)x2 hmz, 2q33.1q37.1(202945934_235502345)x2 hmz	51.5 32.6	parental SNP-array indicated maternal mixUPD2, trio-WES indicated no clinically relevant mutations	Term birth, BW 2.8 kg, healthy
52	Case 83	33 y	17 w+	High risk of T15 detected by NIPT, FGR, anal atresia	1p36.33p35.3(888659_29571758)x2 hmz	28.7	parental SNP-array indicated maternal isoUPD1p36.33p35.3, trio-WES indicated no relevant mutations, CNVseq indicated T15 of placenta	Preterm birth, BW 0.9 kg, anal atresia and heart malformation
53	Case 86	18 y	24 w+	Hypoplasia of the nasal bone, increased echogenicity of fetal bowel	5q15q21.3(96410063_107997118)x2 hmz	11.59	Normal karyotype	Term birth, BW 3.2 kg, healthy
54	Case 87	29 y	17 w+	NIPT indicated a 55.18 Mb deletion at 13q21.1q34	13q21.32q34(66733074_115095705)x2 hmz	48.36	parental SNP-array indicated paternal isoUPD13q21.3q34, trio-WES indicated no relevant mutations, CNVseq indicated mosaic deletion of the placenta in 13q11q34	Term birth, BW 3.6 kg, healthy

GA, gestational age; ROH, region of homozygosity; SNP array, single nucleotide polymorphism array; T, trisomy; FGR, fetal growth restriction; AC, abdomen circumference; FL, femur length; HL, humerus length; /, no further genetic results; IBD, identity by descent; isoUPD, isodisomy; WES, whole-exome sequencing; BW, body weight; y, years old; TOP, termination of pregnancy; NIPT, non-invasive prenatal testing; y, years old; w, weeks.