Cell Guidance Systems Genetics Service
Cytogenetics Laboratory

Customer Name
Customer Name

Laboratory No: SP-XXX
Cell Guidance Reference No: AXXX

Array on XXXX, PX

Date of Receipt:	XXX	Analysed by:	IS
Date of Report:	XXX	Report generated by:	RS

Result Summary
ARRAY REPORT SUMMARY:CONSISTENT WITH A COMPLEX ABNORMAL TRIPLOID KARYOTYPE
ARRAY ISCN: arr[GRCh38]
Xp22.31(7105637_7417150)x1,Xq13.1q22.1(70651742_102996362)x3,Xq22.1q26.2(102996363_132572888)x4,Xq26.2q28(132572889_156040895)x3,1p35.3p35.1(27691007_33687488)x3,1p32.3q41(53529370_222939354)x3,1q41q43(222939355_241468487)x4,1q43(241473155_241612831)x1,1q43q44(241612832_245115611)x4,1q44(245124206_248918679)x1,2q22.1(141047782_141364304)x0,2q22.1(141382080_141490370)x1,2q22.2(141623517_141881744)x1,2q34(211918078_211960274)x0,2q34(211964046_212371756)x1,3p26.3p26.2(1_2849071)x3,3p26.2(2849072_3256437)x1,3p26.2p25.3(3256438_8486744)x3,3p25.3(8486745_9165441)x1,3p25.3p14.2(9175891_59793817)x3,3p14.2(59835694_59993820)x1,3p14.2q13.31(60460206_115019104)x3,3q13.31(116647891_116897946)x1,3q13.31(116898602_117143734)x0,3q13.31(117145824_117205704)x1,3q23q24(142652185_143829313)x3,3q26.32q29(177656243_198136508)x1,4p16.3q35.2(1_190016707)x2
hmz,4p15.31p13(20542194_41602402)x1,4p13p11(44262946_49084990)x1,4q34.3q35.2(180662999_190016707)x1,5q31.1(134054931_134494364)x1,5q34q35.3(163875077_181538259)x3,6p25.3p22.1(1_29442233)x3,6p25.3q27(1_170610382)x2 hmz,6q21q27(109869792_170805979)x3,7p22.3q33(1_134231008)x2
hmz,7q33q36.3(134176009_159345973)x3,8p23.3p12(220692_36077753)x1,9p24.3p22.2(133828_18073938)x1,9p22.2p21.3(18076281_22551241)x3,9q21.31(79167270_79931777)x4,10p15.3q26.3(49904_133620799)x2
hmz,10p12.1p11.1(27054303_38857126)x1,10q21.1(51809621_52826288)x3,13q11q12.13(18445955_26412573)x1,13q21.2q21.32(61754215_68069098)x4,13q21.32q32.1(68069099_94826084)x3,13q32.3q34(100637341_114341521)x2
hmz,13q32.3q34(100637341_114341521)x1,15q11.2q22.31(22033739_65176351)x1,15q11.2q22.31(22033739_65255981)x2
hmz,16p11.2(29681093_31040175)x3,17q11.1q21.2(27052418_41587405)x1,17q21.2q25.3(41587406_83257441)x3,18p11.32p11.21(11358_15286252)x1,18q11.1q12.1(20805338_29926206)x3,18q12.1q12.3(29926207_41114472)x4,18q12.3q23(41114473_80252737)x1,19p13.3q13.43(1_58586566)x2
hmz,19q12(29725443_30146794)x3,20p12.1(14542248_14825931)x1,20p12.1(14879833_15159231)x0,20p12.1(15164442_15298849)x1,20p12.1(15811806_15956779)x1,21q11.2q22.3(13977408_46709983)x3,22q11.1q13.2(15352367_40929469)x3,22q11.1q13.33(15382411_50749012)x2 hmz

Result

DNA from the immortalized cell line, XXXX, PX, was tested for copy number changes and loss of heterozygosity (LOH) using the Illumina Infinium GSA v3 SNP genotyping array.
Array analysis has shown an apparent triploid chromosome constitution with many acquired DNA copy number changes as well as regions of absence of heterozygosity. No Y chromosome was detected. The abnormalities are listed below in chromosome order:
- An ~6Mb gain of 1p35.3p35.1, an ~169Mb gain of 1p32.3q41, an ~18.5Mb high copy number gain of 1q41q43, an ~140kb loss of 1q43 including FH, KMO, OPN3 genes, an ~3.5Mb high copy number gain of 1q43q44, an ~3.8Mb terminal loss of 1q44;
- Three focal losses of 2q22.1 within LRP1B gene, an ~316kb homozygous loss of exons 4-11, an about 108kb heterozygous loss of exon 3 and ~258kb heterozygous loss of exon 2. Further two focal losses of 2q34, an ~42kb homozygous loss of exon 3 in ERBB4 and ~407kb heterozygous loss of exon 2 in ERBB4;
- Gain of the entire short arm of chromosome 3 and the proximal long arm of chromosome 3 from 3p telomere to 3q13.31, this region also contains three focal losses, an ~407kb loss of 3p26.2 including CRBN gene, an ~679k loss of 3p25.3, an ~158kb loss of 3p14.2 including exons 8-9 of the FHIT gene, a compound heterozygous loss of 3q13.31, including an ~245kb homozygous loss, an ~1.2Mb gain of 3q23q24 and an ~20.5Mb terminal loss of

3q26.32q29; - An absence of heterozygosity for the entire chromosome 4 , an ~21Mb interstitial loss of 4p15.31p13, an ~4.8Mb loss of 4p13p11, an ~9.3Mb terminal loss of 4q34.3q35.2;
- An ~439kb interstitial loss of 5q31.1 and an ~17.7Mb terminal gain of 5q34q35.3;
- An absence of heterozygosity for the entire chromosome 6, an ~29.4Mb terminal gain of 6p25.3p22.1 and an ~61Mb terminal gain of 6q21q27;
- An absence of heterozygosity for the entire chromosome 7, an ~25Mb terminal gain of 7q33q36.3; - An ~36Mb terminal loss of 8p23.3p12;
- An ~17.9Mb terminal loss of 9p24.3p22.2, an ~4.5Mb adjacent interstitial gain of 9p22.2p21.3, an ~765kb interstitial high copy number gain of 9q21.31;
- An absence of heterozygosity for the entire chromosome 10, an ~11.8Mb interstitial loss of 10p12.1p11.1 including
ZEB1, an ~1Mb interstitial high copy number gain of 10q21.1 including DKK1 gene;
- An~8Mb proximal loss of 13q11q12.13, an ~6.3Mb high copy number gain 13q21.1q21.32, an ~26.8Mb gain of 13q21.32q32.1, an ~13.7Mb terminal loss of 13q32.2q34 with a complete absence of heterozygosity of the same region;
- An ~43.1Mb proximal loss of 15q11.2q22.31 with a complete absence of heterozygosity;
- An ~1.4Mb interstitial gain of 16p11.2;
- An ~ 14.5Mb loss of 17q11.1q21.2 including NF1 gene, an adjacent ~41.7Mb terminal gain of 17q21.2q25.3; - An ~15.3Mb terminal loss of 18p11.32p11.21, an ~9.1Mb gain of 18q11.1q12.1, an adjacent ~11.2Mb high copy number gain of 18q12.1q12.3, an adjacent ~39.1Mb terminal loss of 18q12.3q23;
- An absence of heterozygosity for the entire chromosome 19, an ~441kb gain of 19q12 including CCNE1 gene; - Four copy number losses of 20q12.1, including an ~279.4 homozygous loss, all within MACROD2 gene; - Gain of the entire chromosome 21;
- An absence of heterozygosity for the entire chromosome 22, an ~25.6Mb gain of 22q11.1q13.2;
- An ~32.3Mb gain of Xq13.1q22.1, an adjacent ~29.6Mb high copy number gain of Xq22.1q26.1, an adjacent ~23.5Mb terminal gain of Xq26.2q28.

Karyotype
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Test Methodology
The analysis was performed in accordance with the published guidelines (Schoumans J et al, 2016. Guidelines for genomic array analysis in acquired haematological neoplastic disorders. Genes, chromosomes and Cancer 55:480-491) using the Nexus Clinical v6.1 software (GRCh 38). The minimum analytical resolution for CNV detection was 5Mb and for LOH detection was 10Mb genome-wide. Balanced chromosomal rearrangements, nucleotide variants, low level clonality (<15%) for any chromosome imbalances, and low level clonality (<20%) for LOH, cannot be excluded using this method.

Remaining DNA from this sample has been destroyed.

Authorized by

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CEO

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Innovative Technology for Regenerative Medicine

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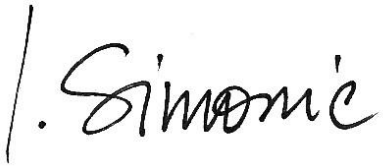

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