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Cytogenomic array detects a subset of myelodysplastic syndrome with increased risk that is invisible to conventional karyotype

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Abstract

Conventional karyotyping is essential standard practice in the initial evaluation of myelodysplastic syndrome (MDS) and is the most impactful single component of the Revised International Prognostic Scoring System (IPSS-R). While single nucleotide polymorphism array (SNP-A) has demonstrated the ability to detect chromosomal defects with greater sensitivity than conventional karyotype, widespread adoption is limited by the unknown additional prognostic impact of SNP-A analysis. Here we investigate the significance of additional SNP-A abnormalities in the setting of MDS and demonstrate differences in survival of patients with additional abnormalities, even those initially characterized as relatively lower risk either by cytogenetic score or IPSS-R. Our findings identify specific abnormalities, particularly *KMT2A* partial tandem duplication (*KMT2A*-PTD), that are invisible to conventional karyotype and potentially contribute to the poor prognosis of MDS patients. Furthermore, these results demonstrate the added value of SNP-A analysis in identifying patients who may benefit from more aggressive therapy, particularly those who would otherwise be classified into lower risk categories.

KEYWORDS: myelodysplastic syndrome, cytogenomic array, SNP array, MDS, SNP-A

INTRODUCTION

Prognostic classification of myelodysplastic syndrome (MDS) relies heavily on cytogenetic abnormalities and is currently utilized to guide therapeutic decision making, including identifying appropriate candidates for bone marrow transplantation. Indeed, cytogenetic risk is the most heavily weighted component of the Revised International Prognostic Scoring System (IPSS-R), which categorizes patients into multiple risk groups of ascending associated poor prognosis.[1] While conventional karyotype is the gold standard for detection of genomic abnormalities in both diagnostic and prognostic settings, single nucleotide polymorphism arrays (SNP-As) have emerged as potential means of further categorizing prognostic risk beyond traditional karyotyping in many hematologic malignancies due to the assay's greater sensitivity in detecting unbalanced chromosomal defects and copy-neutral loss of heterozygosity (CN-LOH).[2-6] However, widespread adoption and incorporation into prognostic algorithms has not yet occurred despite evidence of the clinical significance of SNP-A in combination with already established karyotypic features.[5-8] We therefore sought to refine our understanding of the significance of additional SNP-A abnormalities and their impact on prognosis and ultimately risk of death. In particular, given the high frequency of MDS cases showing a normal karyotype,[9,10] we were especially interested in the potential impact of SNP-A in this group of patients.

MATERIALS AND METHODS

This study was approved by the Institutional Review Board of the University of Michigan. We retrospectively reviewed 108 consecutive patients who underwent karyotyping and Thermo Fisher Cytoscan® array (SNP-A) analysis[11] for diagnosis/classification of a suspected myeloid neoplasm and identified 77 patients with a diagnosis of *de novo* MDS, excluding therapy-related cases. Among these, we identified cases for which additional abnormalities were detected by SNP-A analysis that were not identified using conventional karyotype. We reviewed each patient's electronic medical record including laboratory values at diagnosis (hemoglobin, absolute neutrophil count (ANC), platelet count, and bone marrow blast percentage). We then compared overall survival based on the presence or absence of additional cytogenomic abnormalities detected by SNP-A on groups that were stratified by cytogenetic

risk and IPSS-R score (Figure 1). Overall survival (OS) was calculated from date of diagnosis to date of death, censoring for patients alive at the completion of the study. Patients with both low-risk karyotype (very good-intermediate cytogenetic risk group) and consistent SNP-A results were compared to patients with similar low-risk karyotype but with additional SNP-A abnormalities. These groups were also stratified by the IPSS-R, and survival was compared in patients with and without additional SNP-A abnormalities. Unpaired t, Mann-Whitney, Chi-Square, and Fisher's tests were used as applicable to compare differences in characteristics between groups with and without additional abnormalities. Log-rank (Mantle-Cox) test was used to compare overall survival between the groups.

RESULTS AND DISCUSSION

Of the 77 patients for whom both karyotype and SNP-A were performed (Table 1), 36 cases had additional abnormalities detected by SNP-A (47% of all cases; Table 2; Figure 2). Follow up time ranged from 1.5-85 months (Table 1). Deletions were the most common finding (29 instances), followed by CN-LOH (19 instances). The most prevalent single abnormality detected was *KMT2A(MLL)* partial tandem duplication (*KMT2A*-PTD) (5 cases). Additional abnormalities included cryptic deletions involving *TET2* (3), *RUNX1* (2), and *CUX1* (1). SNP-A detected a monosomy 7 in two cases whose conventional cytogenetics showed a normal karyotype after examining adequate number of metaphase cells.

Cases with and without additional SNP-A abnormalities showed similar overall distribution of morphologic categorization and no statistically significant differences in hemoglobin, ANC, platelet count, bone marrow blast percentage, or IPSS-R (Table 1; Supporting Information Figure S1).

Among matched cases with very good-intermediate cytogenetic risk, those that had additional abnormalities detected on SNP-A showed worse overall survival (median 35.4 months) than those that did not have additional abnormalities (median survival not reached) (Figure 3A; P= 0.010). Similar observations were seen when cases were stratified into matching IPSS-R categories, where very low-intermediate risk cases with additional SNP-A abnormalities showed worse overall survival (median 35.4 months) similar to cases of high-very high risk (median 31.3 months) compared to very low-intermediate risk cases without additional SNP-A abnormalities (median 62.6 months) (Figure 3B; P =0.020).

On the contrary, in cases with either poor-very poor cytogenetic risk or high-very high IPSS-R score, the presence of additional SNP-A abnormalities did not show a statistically significant impact on

overall survival though a trend towards poorer survival was observed (Figures 3A, *P* =0.054 and 3B, *P* =0.052). This finding suggests that the prognostic impact of SNP-A abnormalities may be primarily concentrated in cases that would otherwise be classified as lower risk either by cytogenetic score or IPPS-R.

We questioned whether or not incorporating additional SNP-A findings would impact the calculated IPSS-R. Taking into consideration cases with anomalies that were greater than 5 Mb excluding CN-LOH and small focal deletions and gains, only 2 cases changed IPSS-R (one from very low to intermediate; one from very low to low). In terms of our analysis, therefore, there was no impact on survival.

Based on our previous observation of a small cohort of low risk MDS cases of *KMT2A*-PTD which showed poor overall survival,[12] we questioned whether this single abnormality could be responsible for the survival differences of the cases with additional SNP-A abnormalities. When *KMT2A*-PTD cases were excluded from the analysis, the effects of additional SNP-A abnormalities when stratifying patients according to cytogenetic risk and IPSS-R were somewhat abrogated. A trend towards poorer survival was still noticeable, but no longer statistically significant (Figure 4A; *P* =0.069 and Figure 4B; *P* =0.063). Consequently, *KMT2A*-PTD may be at least partly responsible for the worse survival seen in patients with additional SNP-A abnormalities and otherwise very good-intermediate cytogenetic risk or very low-intermediate IPSS-R, though other abnormalities may also contribute to a lesser extent. Genomic locations of *KMT2A*-PTD in five cases are shown in Figure 5.

To summarize, our findings suggest that the presence of additional SNP-A abnormalities, detected in almost half of MDS cases, has further impact on prognosis and overall survival than that afforded by conventional karyotype analysis. Cases identified as very good-intermediate cytogenetic risk that have additional SNP-A abnormalities demonstrate overall survival approaching that of patients with poor-very poor cytogenetic risk. Similarly, cases identified as very low-intermediate risk by IPSS-R demonstrate overall survival more similar to patients with high-very high risk by IPSS-R. Although a significant component of these differences may be accounted for by the presence of *KMT2A*-PTD, the data imply that other additional abnormalities also impact the prognosis of MDS.

Although the types of abnormalities we noted in our study were largely similar to those reported in previous studies analyzing SNP-A in MDS patients,[4-7] the frequency of additional SNP-A abnormalities in our study of 47% was higher than previous reports, which showed a range of 18-39% of

cases with additional SNP-A abnormalities. The increased frequency we observed could potentially be due to higher resolution of our array platform and as well as our analysis. Coincident with this observation, *KMT2A*-PTD was only rarely reported in these prior studies, whereas in our study this abnormality was the most common additional SNP-A abnormality observed.

The data also demonstrate that the effect of additional SNP-A abnormalities is particularly profound in cases that have very good-intermediate karyotypic features or cases of otherwise low-intermediate risk by IPSS-R. Thus, the SNP-A may be more effectively utilized in these particular cases and, perhaps not surprisingly, is more limited in usefulness in cases that already have definitive poor risk features. Moreover, because a large proportion of cases in the very low-intermediate risk/very good-intermediate cytogenetic groups are patients with a normal karyotype, SNP-A is a potentially useful tool in further delineating risk subgroups within this substantial fraction of MDS cases. Interestingly, the survival of patients in our study was longer than those of prior studies that performed survival analysis,[5,7] which showed a median survival of 43-50 months in patients with favorable or normal karyotype without SNP-A abnormalities and 16-20 months in patients with additional SNP-A abnormalities. The relatively increased overall survival seen in our cohort compared to prior studies may potentially be attributed to either differences in patient population, clinical practice, or a consequence of relatively smaller cohort size.

The contribution of individual specific abnormalities to prognosis also remains an area of future investigation. Our data suggest *KMT2A*-PTD may be partially responsible for the poorer survival seen in MDS patients, who otherwise might be classified as having lower risk disease, though a definitive determination is limited by the small number of *KMT2A*-PTD. Additionally, two cases also identified CN-LOH as additional anomalies on chromosome arms 7q and 11q, respectively, which contain the genes *EZH2* on 7q and *CBL* on 11q, both recurrently mutated in MDS with suggested adverse prognostic impact.[13-16] Consequently, the relative contribution of *KMT2A*-PTD to prognosis requires further confirmation in larger sample study. Because patients with additional SNP-A abnormalities have poorer overall survival and relatively increased risk, the presence of these abnormalities may potentially identify these patients as candidates for more urgent therapeutic intervention including transplantation.

While conventional karyotype continues as expected standard practice in MDS diagnosis, the appropriate utilization of other related ancillary testing to provide a comprehensive genetic assessment is an important as-yet unresolved issue. For example, fluorescence in situ hybridization (FISH) analysis can

also detect additional abnormalities outside of karyotype,[17,18] however the National Comprehensive Cancer Network (NCCN) practice guidelines currently only recommend FISH in cases where standard cytogenetics cannot be obtained.[19] The American Society for Clinical Pathology also supports a similar recommendation.[20] The European Society for Medical Oncology (ESMO) practice guidelines slightly differ in this respect as they acknowledge a potential benefit of FISH in cytogenetically normal cases and thus recommend FISH in the setting of normal karyotype.[21] A similar algorithm could potentially be applied with respect to SNP-A by focusing its utilization in cases that would otherwise be designated of lower risk. Recent studies have attempted to compare the relative detection rates of abnormalities across different modalities and have confirmed overall relatively higher resolution in SNP-A[22] and next generation sequencing (NGS) platforms which complement standard cytogenetics and FISH analysis.[23] Outside of larger genomic alterations, the impact of point mutations, which have also been implicated in survival of MDS patients with further prognostic prognostic relevance,[15,24] remains another area for subsequent investigation. As clinical standard of care does not yet include testing for point mutations, we were unable to ascertain their significance in the context of additional SNP-A abnormalities, but this is an area in which further analysis is warranted.

In conclusion, these findings demonstrate the potential prognostic and therapeutic impact of the cytogenomic array, with particular utility in MDS cases that would otherwise be classified as very low-intermediate risk.

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Figure Legends

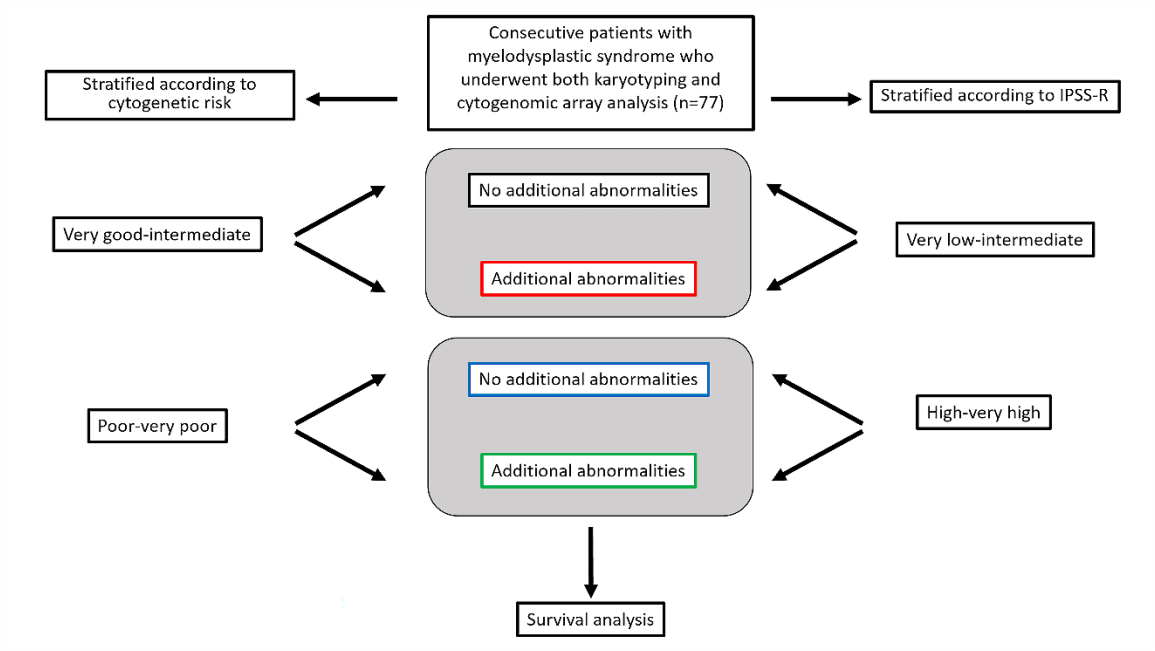
FIGURE 1 Study design. MDS cases for which both karyotyping and cytogenomic array (SNP-A) was performed were stratified by 1) cytogenetic risk and 2) Revised International Prognostic Scoring System (IPSS-R). They were then further stratified based on the presence or absence of additional SNP-A abnormalities. Survival analysis was performed.

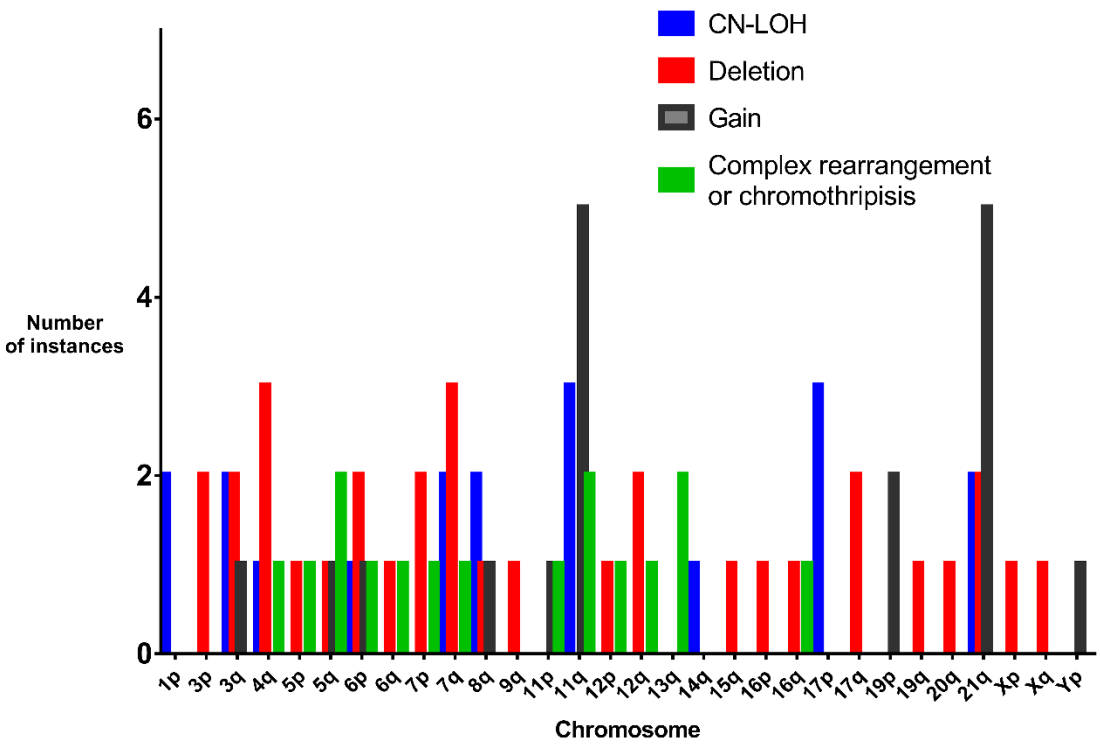
FIGURE 2 Frequency of additional SNP-A abnormalities. The number of instances of each type of additional abnormality, which were not detected by conventional karyotype, are depicted for each chromosome.

FIGURE 3 The presence of additional SNP-A abnormalities negatively impacts survival in patients with very good-intermediate cytogenetic risk and very low-intermediate IPSS-R. Survival analysis of patients stratified by cytogenetic risk (A; $P<0.0001$) and by IPSS-R (B; $P=0.002$) was performed comparing cases with and without additional SNP-A abnormalities. (A) Cases with very good-intermediate cytogenetic risk with additional SNP-A abnormalities showed worse overall survival than those that did not have additional abnormalities ($P = 0.010$; black and red groups). SNP-A abnormalities had no statistically significant survival impact in cases with poor-very poor cytogenetic risk ($P=0.054$; green and blue groups). (B) Very low-intermediate IPSS-R cases with additional SNP-A abnormalities showed worse overall survival compared to those without additional abnormalities ($P=0.020$; black and red groups). SNP-A abnormalities had no statistically significant survival impact in cases with high-very high IPSS-R score ($P=0.052$; green and blue groups).

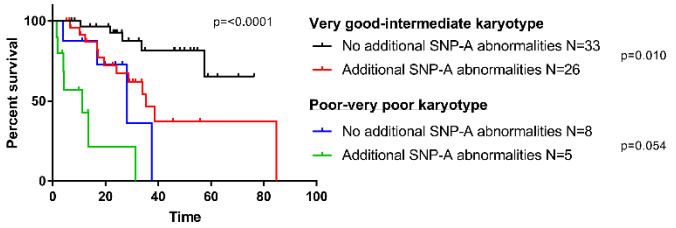
FIGURE 4 *KMT2A*-PTD significantly impacts survival of patients with additional SNP-A abnormalities. (A) Patients with very good-intermediate karyotype showed worse survival in the presence of additional SNP-A abnormalities (P =0.010), an effect which was abrogated but not completely eliminated when cases of *KMT2A*-PTD were excluded (P =0.069). (B) Similar survival effects were seen in cases with very low-intermediate IPSS-R with (P =0.020) and without (P =0.063) *KMT2A*-PTD.

FIGURE 5 Genomic locations and graphic display of array results for *KMT2A*-PTD. Patient 7 had gain of 11q23.3 at the other homologue of chromosome 11.

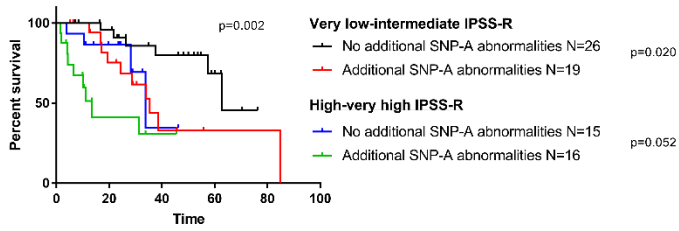


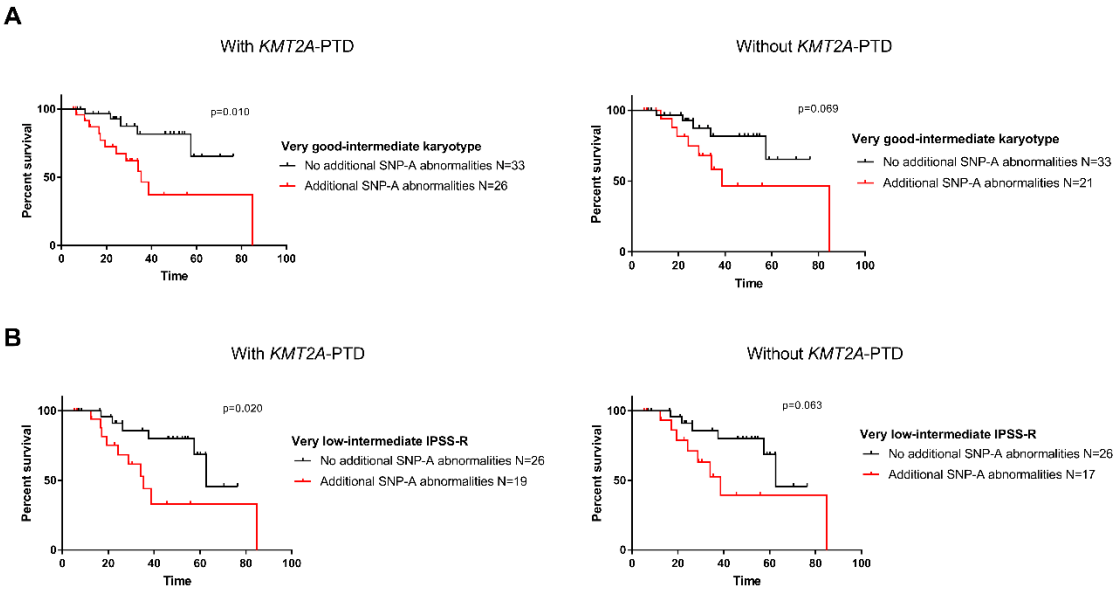


A Stratified by cytogenetic risk



B Stratified by IPSS-R





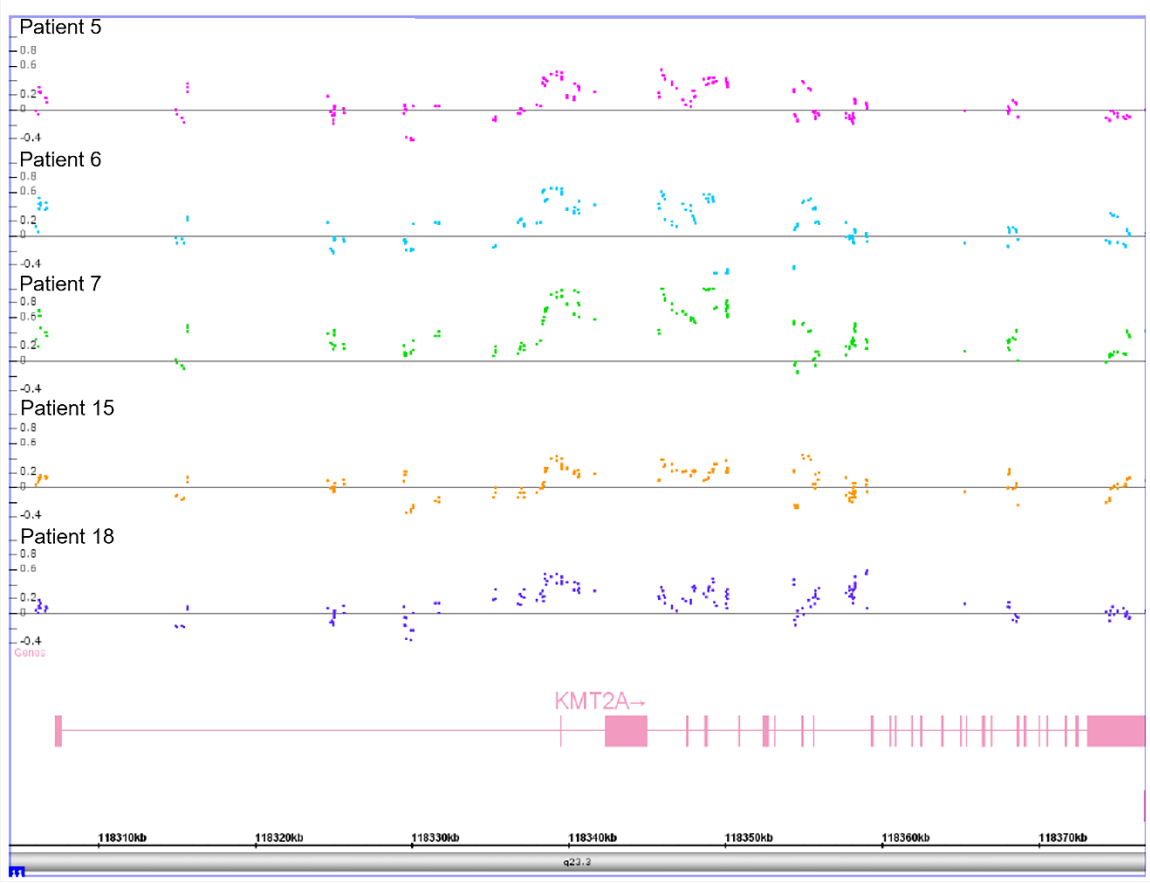


TABLE 1 Clinical characteristics of study group

		Cytogenetic risk group					
		Very good-intermediate			Poor-very poor		
	Total	No additional SNP-A abnormalities	Additional SNP-A abnormalities	*P*	No additional SNP-A abnormalities	Additional SNP-A abnormalities	*P*
Number of patients	77	33	26		8	10	
Age		66 (59-74)	71 (64-78)	*0.18*	64.4 (60.6-75.5)	71.2 (62.9-80.0)	*0.36*
Sex	77			*0.59*			*0.64*
Male	50	20	18		6	6	
Female	27	13	8		2	4	
Initial diagnosis	77			*0.29*			*0.46*
MDS-SLD	8	3	4		1	0	
MDS-MLD	35	17	10		4	4	
MDS-EB1	12	6	2		2	2	
MDS-EB2	22	7	10		1	4	
IPSS-R		3 (2.5-4.9)	2.75 (1.5-4.25)	*0.16*	6 (4.25-7.6)	7.25 (6-8.5)	*0.14*
ANC, k/uL		1.6 (0.8-3.4)	1.7 (1.0-2.5)	*0.79*	0.9 (0.3-2.3)	0.8 (0.5-1.4)	*0.85*
Hgb, g/dL		9.8 (8.1-11.5)	9.6 (8.0-10.9)	*0.73*	9.0 (7.6-9.9)	9.1 (8.0-9.5)	*0.56*
Plt, k/uL		75.5 (53-191)	118 (66-180)	*0.6*	65 (30-110)	43 (18-59)	*0.37*
BM blast percent, %		2.3 (0.9-6.1)	2.0 (1.0-4.3)	*0.95*	1.8 (0.3-6.7)	3.4 (1.3-9.8)	*0.24*
Therapy				*0.8*			*NA*
Supportive		15	12		2	4	
Hypomethylating agent		18	12		5	7	
Transplant		9	5		3	0	
Other		5	6		0	0	
Median follow up time, months		32.6	26.5		21.7	7.2	

		IPSS-R					
		Very low-intermediate risk			High-very high		
		No additional SNP-A abnormalities	Additional SNP-A abnormalities	*P*	No additional SNP-A abnormalities	Additional SNP-A abnormalities	*P*
Number of patients	77	26	20		18	13	
Age		67.9 (59.7-75.0)	74.0 (62.8-79.8)	*0.22*	62.5 (59.5-71.4)	69.1 (65.4-76.3)	*0.14*
Sex	77			*0.35*			*>0.99*
Male	50	15	15		12	8	
Female	27	11	5		6	5	
Initial diagnosis	77			*0.16*			*0.69*

MDS-SLD	8	5	3		0	0	
MDS-MLD	35	16	11		4	4	
MDS-EB1	12	4	1		5	2	
MDS-EB2	22	1	5		9	7	
IPSS-R		2.75 (2-3.5)	2 (1.5-3)	*0.3*	6 (5.5-6.5)	6.25 (5.6-7.9)	*0.3*
ANC, k/uL		1.6 (0.8-3.3)	2 (1.1-2.6)	*0.37*	1.2 (0.4-3.1)	0.9 (0.5-1.9)	*0.83*
Hgb, g/dL		10.1 (8.6-12.3)	10.1 (8.1-11.3)	*0.66*	8.3 (7.5-9.8)	9.0 (7.6-9.4)	*0.86*
Plt, k/uL		87.5 (69.5-164)	118 (69.8-189)	*0.62*	44 (28-81)	45 (24-122)	*0.98*
BM blast percent, %		1.6 (0.6-2.5)	1.9 (0.8-2.1)	*0.77*	8 (1.5-10)	7.4 (2.1-12)	*0.58*
Therapy				*0.99*			*0.19*
Supportive		14	10		3	6	
Hypomethylating agent		13	9		10	10	
Transplant		5	3		7	2	
Other		4	3		1	3	
Median follow up time, months		46.2	26.5		24.2	10.3	

Median value (25th-75th percentile) displayed; absolute neutrophil count (ANC), hemoglobin (Hgb), platelet count (Plt), bone marrow (BM), NA (unable to perform), "Other" therapy (therapies not listed above)

TABLE 2 Additional abnormalities detected by cytogenomic array

				SNP-A results			S
Patient	**Karyotype**	**Cytogenetic risk**	**Cytogenetic risk after array (anomalies >5 Mb but excluding CN-LOH and small focal deletions, duplications)**	**Array results**	**%**	**Size (Mb)**	**S c w k**
1	46,XY[20]	Good	Good	arr[hg19] 4q11q35.2(52,686,799-190,921,709)x2 hmz	90	138.2	
2	47,XXX?c[20]	Good	Good	arr[hg19] 21q11.2q22.3(14,386,012-48,084,820)x2 hmz	30-90	33.7	
				arr[hg19] Xp22.33q28(168,546-155,233,731)x3	100	155.1	Y
3	46,XY[16]	Good	Intermediate	arr[hg19] 15q14q22.2(35,437,654-61,211,671)x1-2	15	25.8	
4	46,XY[20]	Good	Poor	arr[hg19] 6q23.2q23.3(134,365,000-136,607,455)x1	100	2.2	
				arr[hg19] 7p22.3q36.3(43,360-159,119,707)x1-2	7	159.1	
				arr[hg19] 11q13.1q25(65,577,515-134,942,626)x2 hmz	100	69.4	
5	46,XX[20]	Good	Good	arr[hg19] 7q35q36.3(144,958,661-159,119,220)x2 hmz	90	14.2	
				arr[hg19] 11q23.3(118,338,293-118,354,345)x2-3	45	16 kb	
6	46,XY[20]	Good	Good	arr[hg19] 11q12.2q25(60,804,709-134,942,626)x2 hmz	90	74.1	
				arr[hg19] 11q23.3(118,338,293-118,349,247)x2-3	>50	11 kb	
7	46,XX[20]	Good	Good	arr[hg19] 11q23.3(118,123,516-118,470,527)x2-4	80	347 kb	
				arr[hg19] 11q24.3(128,408,210-128,699,707)x2-3	80	291 kb	
8	46,XY[20]	Good	Good	arr[hg19] 1p36.33p34.1(882,802-45,000,436)x2 hmz	90	44.1	
9	46,XY[20]	Good	Good	arr[hg19] 3q11.1q29(93,735,022-197,851,260)x2 hmz	35	104.1	
				arr[hg19] 4q24(106,130,009-106,190,922)x1-2	50	61 kb	
10	46,XX[20]	Good	Good	arr[hg19] 7q21.3q36.3(97,735,123-159,119,220)x2 hmz	50	61.4	
				arr[hg19] 21q11.2q22.3(15,867,134-48,084,820)x2 hmz	50	32.2	
11	46,XX[20]	Good	Good	arr[hg19] 8q11.23q24.3(53,704,149-146,292,734)x2 hmz	15	92.6	
12	46,XY[20]	Good	Good	arr[hg19] 4q22.1q35.2(92,145,040-190,921,709) x2 hmz	10	98.8	
13	46,XY[20]	Good	Poor	arr[hg19] 7p22.3q36.3(43,360-159,119,707)x1-2	12	159.1	
14	46,XY[20]	Good	Good	arr[hg19] 3q21.3q29(126,531,213-197,851,260)x2 hmz	85	71.3	
				arr[hg19] 7q22.1(99,829,321-102,058,793)x1-2	80	2.2	
				arr[hg19] 21q22.12(36,294,421-37,432,271)x1-2	85	1.1	
15	46,XX[20]	Good	Good	arr[hg19] 11q23.3(118,338,521-118,355,688)x2-3	30	17 kb	
16	46,XY[20]	Good	Good	arr[hg19] 4q24(105,995,910-106,227,999)x1-2	90	0.2	
17	46,XY[20]	Good	Good	arr[hg19] 21q22.12(36,002,849-37,408,933)x1-2	80	1.4	

18	46,XY[20]	Good	Good	arr[hg19] 9q34.3(139,101,277-139,734,766)x1	80	0.6	
				arr[hg19] 11q23.3(118,335,185-118,359,052)x3	80	24 kb	
19	46,XY[20], NUP98 rearrangement (80% by FISH)	Good	Good	arr[hg19] 5q35.3(176,650,787-176,768,901)x2-3	80	0.1	
				arr[hg19] 11p15.4(3,764,205-3,832,210)x2-3	80	68 kb	
20	46,XY,del(5)(q15q33)[4]/46,XY[1]	Good	Good	arr[hg19] 1p36.33p31.3(903,425-65,250,982)x2 hmz	60	64.3	
				arr[hg19] 5q15q33.2(93,274,523-154,360,732)x1-2	40	64.1	Y
21	46,XY,del(5)(q15q31)[19]/46,XY[1]	Good	Good	arr[hg19] 3p13p11.1(70,310,611-88,552,092)x1-2	60	18.2	
				arr[hg19] 5q15q31.1(95,982,582-131,916,380)x1-2	70	35.9	Y
22	46,XX,del(5)(q13q33)[12]/46,XX[8]	Good	Good	arr[hg19] 5q14.2q34(81,856,536-160,672,001)x1-2	50	78.8	Y
				arr[hg19] 12p13.31p13.1(10,000,550-13,258,017)x1-2	30	3.3	
24	46,XY,del(13)(q12q14)[7]/46,XY[18]	Intermediate	Intermediate	arr[hg19] 4q24(105,942,532-106,564,759)x1-2	95	0.6	
				arr[hg19] 13q13.1q14.3(33,109,828-53,700,736)x1-2	25	20.6	Y
				arr[hg19] Xq28 or Yq12(154,941,868-155,233,731 or 59,044,874-59,336,737)x1-2	95	0.3	
26	47,XY,+8[18]/46,XY[2]	Intermediate	Intermediate	arr[hg19] 8p23.3q24.3(158,048-146,295,771)x2-3	30	146.1	Y
				arr[hg19] 11q13.2q25(67,015,468-134,942,626)x2 hmz	50	69.9	
27	47,XX,+8[14]/46,XX[6]	Intermediate	Intermediate	arr[hg19] 3q26.2(168,582,060-170,230,667)x2-3	90	1.6	
				arr[hg19] 8p23.3q11.1(158,048-47,126,524)x2-3	65	47	
				arr[hg19] 8q11.1q11.21(47,127,862-51,456,633)x1-2	90	4.3	
				arr[hg19] 8q11.21q24.3(51,456,754-146,295,771)x2-3	65	94.8	
29	44,XX,add(4)(q23),-5,+6,der(6;12)t(6;12)(p21;p11.2)del(6)(p12p21),der(6)t(6;13)(p21;q14)del(6)(p12p21),der(11)dup(11)(q13q13)del(11)(q23q23),-13,add(14)(p11.2),del(18)(q21q23)[cp13]/45,sl,+mar[7]	Very poor	Very poor	arr[hg19] 4q25(109,354,361-111,191,990)x1	100	1.8	
				arr[hg19] 4q32.3q35.2(167,790,246-190,957,473)x1	100	23.2	Y
				arr[hg19] 5p15.33p15.2(113,576-13,611,558)x1	100	13.5	
				arr[hg19] 5q11.2q35.1(53,519,660-171,907,198)x1	100	118.4	
				arr[hg19] 6p24.1p22.1(11,797,999-27,746,178)x1	100	15.9	
				arr[hg19] 6p22.1p21.1(30,105,444-44,326,337)x3	100	14.2	Y
				arr[hg19] 6p21.1p12.1(44,326,504-56,764,165)x1	100	12.4	Y
				arr[hg19] 6p12.1q27(56,768,218-170,919,482)x3	100	114.2	Y
				arr[hg19] 11p11.12q25(51,126,723-134,938,470)cx	100	83.8	
				arr[hg19] 12p13.2p11.21(11,658,944-31,485,751)x1	100	19.8	Y
				arr[hg19] 13q11q34(19,436,286-115,107,733)cx	100	95.7	
				arr[hg19] 17q22(54,260,365-55,515,281)x1	100	1.3	
				arr[hg19] 18q21.2q23(52,763,256-78,014,123)x1	100	25.3	Y
30	43,XY,der(3)t(3;16)(p12;q	Very poor	Very poor	arr[hg19] 3p21.2p12.2(50,699,382-83,166,160)x1-2	65	32.5	Y

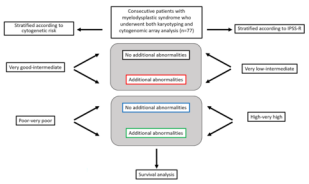
	13),der(5)t(3;5)(p12;q13),-12,der(13)t(12;13)(q12;q34),der(17)t(17;20)(p13;p11.2),-20,idic(22)(p11.2)[cp18]/46,XY[2]			arr[hg19] 3p11.2q22.3(87,417,120-136,702,414)x1-2	10	49.3	
				arr[hg19] 5q13.2q35.3(72,110,523-180,719,789)x1-2	65	108.6	Y
				arr[hg19] 8q24.13q24.21(126,229,398-130,825,360)x2-3	65	4.6	
				arr[hg19] 12p13.33q12(173,786-46,058,033)x1-2	65	45.9	Y
				arr[hg19] 12q13.11q13.12(48,829,668-49,813,922)x1-2	65	1	
				arr[hg19] 12q21.1q21.2(75,107,892-76,608,109)x1-2	65	1.5	
				arr[hg19] 14q11.2q32.33(20,511,672-107,285,437)x2 hmz	90	86.8	
				arr[hg19] 16p11.2(28,689,085-32,922,512)x1-2	65	4.2	
				arr[hg19] 17p13.3p13.1(9,474-8,172,907)x1-2	65	8.2	Y
				arr[hg19] 19q13.32q13.33(47,126,613-49,500,959)x1-2	65	2.4	
				arr[hg19] 20p12.1(15,681,353-17,022,497)x1-2	65	1.4	
				arr[hg19] 20p11.21q11.21(22,735,537-29,871,042)x1-2	65	7.1	
				arr[hg19] 20q11.21(29,874,663-31,364,166)x3	100	1.5	
				arr[hg19] 20q11.21q13.33(31,382,491-62,897,159)x1-2	65	31.5	
				arr[hg19] 22q11.1q13.33(16,888,899-51,197,838)x2-3	55	34.3	Y
31	47,XY,t(1;3)(p13;q21),del(4)(q21q25),del(11)(q14q24),+19,del(20)(q11.2q13.3)[4]	Very poor	Very poor	arr[hg19] 4q21.23q25(84,749,459-109,483,856)x1-2	60	24.7	Y
				arr[hg19] 11q14.2q24.1(86,711,530-123,487,591)x1-2	60	36.8	Y
				arr[hg19] 17q11.2(29,262,000-30,466,769)x1-2	60	1.2	
				arr[hg19] 19p13.3q13.43(260,911-58,956,888)x2-3	20	58.7	Y
				arr[hg19] 20q11.21q13.31(31,062,502-56,290,652)x1-2	50	25.2	Y
				arr[hg19] 21q22.12(36,202,439-36,282,500)x2-3	100	80 kb	
				arr[hg19] Xp22.2(15,644,219-16,702,011)x1-2	50	1.1	
32	44-46,XY,der(2)ins(2;6)(q23;p24p12)add(2)(q23),der(3)t(3;12)(p24;p13),add(4)(q12),der(5;22)(p10;q10),-6,der(12)t(3;12)t(?6;12)(q12;q24),der(19)dup(19)(q13.1q13.4)add(19)(q13.4),+mar[cp17]/43-44,sl,add(X)(p11.2),-der(2)ins(2;6)add(2),+add(2)(q32),-der(3)t(3;12),+3,+6,-7,-der(12)t(3;12)t(6;12),+add(12)(p13),add(19)(p13),add(?21)(p11.2),+del(?22)(q11.2q13),-mar[cp3]	Very poor	Very poor	arr[hg19] 3p26.3p12.3(61,891-74,491,151)cx	80	74.4	Y
				arr[hg19] 4q12q32.2(57,131,170-163,425,170)cth	80	106.3	
				arr[hg19] 5q11.2q12.1(54,136,001-60,742,205)x1-2	90	6.6	Y
				arr[hg19] 5q12.1q35.3(61,997,673-177,260,317)x1-2	90	115.3	Y
				arr[hg19] 6p25.3p24.2(156,974-10,880,159)x1-2	80	10.7	
				arr[hg19] 16q22.3q23.2(73,602,589-80,595,329)x1-2	80	7	
				arr[hg19] 17p13.3p11.2(18,900-16,402,114)x2 hmz	90	16.4	
				arr[hg19] 19q13.11q13.43(34,783,132-58,956,888)x2-3	80	24.2	Y
				arr[hg19] 20q11.23q13.32(34,882,014-58,360,984)x1-2	20	23.5	

				arr[hg19] 21q22.13q22.3(39,246,697-43,050,829)x2-3	30	3.8	
				arr[hg19] 22q11.1q13.1(16,888,899-38,450,184)x2-3	80	21.6	Y
				arr[hg19] 22q13.1q13.33(39,235,339-51,197,838)x2-3	80	12	Y
33	44,XY,t(1;12)(q21;q24.1),-3,add(3)(p11),der(5)t(3;5)(p13;q23),-6,-7,+11,der(11)t(3;11)(q21;q13)[15]/46,XY[5]	Very poor	Very poor	arr[hg19] 1q21.3q22(153,368,019-155,017,913)x1-2	30	1.7	Y
				arr[hg19] 3p26.1q29(4,103,600-197,851,936)cx	30	193.8	Y
				arr[hg19] 5q23.2q35.3(124,723,524-180,719,789)x1-2	30	56	Y
				arr[hg19] 6p25.3p22.3(156,974-24,311,197)x1-2	30	24.2	Y
				arr[hg19] 6p22.2p22.1(26,553,570-28,222,528)x1-2	30	1.7	Y
				arr[hg19] 6p21.1p12.3(43,395,395-47,462,571)x2-3	30	4.1	
				arr[hg19] 6p12.3q27(47,467,934-170,919,482)x1-2	30	123.5	
				arr[hg19] 7p21.3p21.1(8,299,324-16,722,353)x1-2	30	8.4	
				arr[hg19] 7p13p12.1(44,817,005-50,784,997)x1-2	30	6	
				arr[hg19] 7q11.21(63,083,343-66,898,842)x1-2	30	3.8	
				arr[hg19] 7q35q36.3(144,075,389-159,119,707)x1-2	30	15	
				arr[hg19] 17p13.3p11.2(18,900-17,027,255)x2 hmz	40	17	
57	46,X,del(Y)(q11.23)[8]/46,XY[12]	Good	Good	arr[hg19] Yp11.31q11.221(2,650,140-19,576,531)x1-2	40	16.9	
				arr[hg19] Yq11.221q11.23(19,585,828-28,799,937)x0-1	40	8.7	
69	45-46,XY,dic(3;5)(5pter->5q11.2::3p12->3q29::3p22->3pter),del(7)(q21q36),+0-1mar[cp18]/46,XY[2]	Very poor	Very poor	arr[hg19] 3p22.2p12.1(37,299,213-83,644,799)x1-2	70	46.4	Y
				arr[hg19] 5q11.1q12.3(49,430,268-64,262,486)x1-2	20-55	14.8	Y
				arr[hg19] 5q12.3q13.3(66,243,349-76,743,838)x1-2	25-70	10.5	Y
				arr[hg19] 5q14.3q35.3(85,913,148-180,719,789)x1-2	70	94.8	Y
				arr[hg19] 7q21.13q36.3(88,745,758-159,119,707)x1-2	70	70.4	Y
70	46~49,XX,-2,del(5)(q15q33),-6,del(6)(p23p24),-7,-13,-15,+16,del(16)(q12q24),+17,add(17)(p11.2),add(17)(p12),-18,-19,-21,+1~2r,+3~5mar[cp10]/46,XX[10]	Very poor	Very poor	arr[hg19] 2q33.1q37.3(201,635,517-242,783,384)x1-2	15	41.2	Y
				arr[hg19] 5q13.3q35.3(73,921,819-180,719,789)x1-2	15	106.8	Y
				arr[hg19] 6p22.3p22.1(18,309,604-28,848,258)x4-6	amp	10.5	
				arr[hg19] 13q11q34(19,436,286-115,107,733)x1-2	15	95.7	Y
				arr[hg19] 19p13.2(10,856,592-12,040,283)x4-7	amp	1.2	
				arr[hg19] 19p13.2p13.12(13,477,560-15,274,712)x2-5	amp	1.8	
				arr[hg19] 21q21.1(19,629,216-20,241,815)x4-7	amp	0.6	
				arr[hg19] 21q22.12q22.3(35,848,786-48,097,372)x2-3	40	12.2	
73	45,XX,der(4)t(4;?13)(q35;q14),-13,-16,der(17)t(?16;17)(p11.2;	Very poor	Very poor	arr[hg19] 1p36.33p32.1(849,466-59,863,870)x2-3	80	59	Y

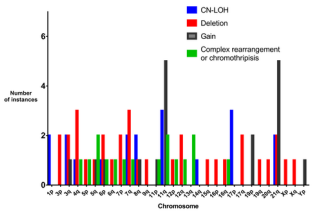
	p11.2),+der(?)t(?;1)(?;p31)[18]/44-45,XX,-8,dic(8;10)(q26;q2?2)ins(10;1)(q26;p31p36),-16,der(17)t(?16;17)[cp2]			arr[hg19] 1p31.1(71,990,325-73,466,675)x2-3	85	1.5	Y
				arr[hg19] 4q35.2(190,712,389-190,957,473)x1-2	85	0.3	Y
				arr[hg19] 8q23.1q24.3(108,384,827-146,292,734)x2 hmz	85	37.9	
				arr[hg19] 13q21.31q21.33(65,565,962-69,826,807)x1-2	85	4.3	
				arr[hg19] 13q33.1(103,508,960-103,531,681)x1-2	85	23 kb	
				arr[hg19] 13q33.1(103,592,015-104,256,190)x1-2	85	0.7	
				arr[hg19] 13q33.2q33.3(105,264,673-109,888,382)x1-2	85	4.6	
				arr[hg19] 16q11.2q24.3(46,503,572-90,155,062)x1	100	43.7	Y
				arr[hg19] 17p13.3p11.2(525-17,988,254)x1	95	18	Y
74	45,XY,del(5)(q22q33),der(17;20)(q10;p10)[3]/46,sl,del(7)(q11.2q36),+8[5]/44,sl,-7[6]/85-94,slx2,add(11)(q12)x2,+1-2mar,4-11dmin[cp4]/46,XY[2]	Very poor	Very poor	arr[hg19] 5q21.3q33.3(106,203,054-157,533,436)x1-2	30	51.3	Y
				arr[hg19] 7p22.3q36.3(43,360-159,119,707)x1-2	15	159.1	Y
				arr[hg19] 8p23.3q24.3(158,048-146,295,771)x2-3	10	146	Y
				arr[hg19] 11q13.4(70,735,084-74,545,922)x1-2	10	3.8	
				arr[hg19] 11q13.4q14.1(74,950,287-80,597,434)x2-3	10	5.7	
				arr[hg19] 11q14.1q14.3(80,885,537-92,686,784)x1-2	10	11.8	
				arr[hg19] 11q22.1q22.3(97,942,172-103,313,943)x1-2	10	5.4	
				arr[hg19] 11q22.3q23.3(104,772,138-117,129,522)x1-2	10	12.4	
				arr[hg19] 11q23.3(117,940,196-118,570,397)x2-3	80	0.6	
				arr[hg19] 11q23.3q24.2(118,719,566-126,066,194)x1-2	10	7.4	
				arr[hg19] 11q24.2q25(126,201,307-134,938,470)x3-5	amp	8.7	
				arr[hg19] 17p13.3p11.2(525-21,722,139)x1-2	30	21.7	Y
				arr[hg19] 20q11.22q13.33(32,359,017-62,915,555)x1-2	30	32.6	Y
				arr[hg19] 21q21.3q22.3(30,253,288-48,097,372)x2-3	10	17.8	
76	46-48,XX,del(1)(p22p36.1),del(2)(p24),del(3)(p21),-5,der(6)t(1;6)(p13;q25),der(7)add(7)(p21)del(7)(q32q36),+8,+8,+8,del(11)(q22q23),-12,+13,del(13)(q12q14)x2,add(16)(q11.2),add(17)(p13),add(19)(p13),-21, +0-2mar[cp20]	Very poor	Very poor	arr[hg19] 1p21.1p13.3(103,620,493-108,243,851)x1-2	70	4.6	Y
				arr[hg19] 3q26.33(180,626,349-180,728,459)x1-2	70	0.1	
				arr[hg19] 5p15.33p14.1(113,576-24,705,254)x1-2	80	24.6	
				arr[hg19] 5p14.1p13.2(26,602,764-36,629,258)x1-2	80	10	
				arr[hg19] 5q14.3(86,075,402-91,536,195)x1-2	80	5.5	
				arr[hg19] 5q21.1q32(99,149,791-149,316,740)x1-2	80	50.2	
				arr[hg19] 5q33.1q34(151,060,202-163,263,177)x1-2	80	12.2	
				arr[hg19] 6q11.1q13(62,118,669-73,010,280)x1-2	80	10.9	Y
				arr[hg19] 6q24.2q25.1(144,387,634-152,252,413)x1-2	80	7.9	Y
				arr[hg19] 7p21.3p21.1(11,371,004-17,035,173)x1-2	80	5.7	Y
				arr[hg19] 7p14.1(37,771,693-40,371,298)x1-2	80	2.6	Y

				arr[hg19] 7q33q36.3(134,801,961-159,119,707)x1-2	80	24.3	Y
				arr[hg19] 8p23.3q24.3(158,048-146,295,771)x3-4	pentasomy	146.1	Y
				arr[hg19] 12p12.3q22(15,277,112-95,085,949)cx	80	79.8	
				arr[hg19] 13q11q13.1(19,436,286-32,565,539)x2-3	70	13.1	Y
				arr[hg19] 13q13.1q21.31(32,576,910-64,735,038)x1-2	70	32.2	Y
				arr[hg19] 13q21.31q34(64,738,525-115,107,733)x2-3	70	50.4	Y
				arr[hg19] 16q12.1(48,341,743-50,857,833)x1-2	80	0.4	
				arr[hg19] 16q12.1q24.3(51,570,940-90,155,062)x1-2	80	2.5	
				arr[hg19] 16q11.2q12.1(46,580,413-47,022,778)x1-2	80	38.6	
				arr[hg19] 17p13.3p11.2(18,900-20,697,797)x2 hmz	80	20.7	

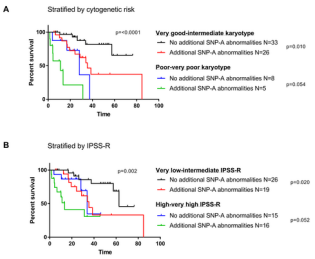
Copy neutral loss of heterozygosity (CN-LOH), partial tandem duplication (PTD), amplification (amp)



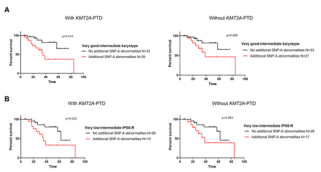
GCC_22783_Figure 1.tif



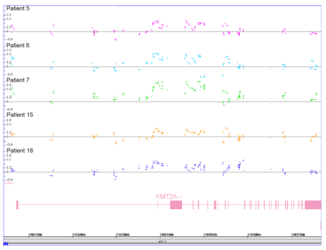
GCC_22783_Figure 2.tif



GCC_22783_Figure 3_2.tif



GCC_22783_Figure 4.tif



GCC_22783_Figure 5.tif

TABLE 1 Clinical characteristics of study group

		Cytogenetic risk group					
		Very good-intermediate			Poor-very poor		
	Total	No additional SNP-A abnormalities	Additional SNP-A abnormalities	*P*	No additional SNP-A abnormalities	Additional SNP-A abnormalities	*P*
Number of patients	77	33	26		8	10	
Age		66 (59-74)	71 (64-78)	*0.18*	64.4 (60.6-75.5)	71.2 (62.9-80.0)	*0.36*
Sex	77			*0.59*			*0.64*
Male	50	20	18		6	6	
Female	27	13	8		2	4	
Initial diagnosis	77			*0.29*			*0.46*
MDS-SLD	8	3	4		1	0	
MDS-MLD	35	17	10		4	4	
MDS-EB1	12	6	2		2	2	
MDS-EB2	22	7	10		1	4	
IPSS-R		3 (2.5-4.9)	2.75 (1.5-4.25)	*0.16*	6 (4.25-7.6)	7.25 (6-8.5)	*0.14*
ANC, k/uL		1.6 (0.8-3.4)	1.7 (1.0-2.5)	*0.79*	0.9 (0.3-2.3)	0.8 (0.5-1.4)	*0.85*
Hgb, g/dL		9.8 (8.1-11.5)	9.6 (8.0-10.9)	*0.73*	9.0 (7.6-9.9)	9.1 (8.0-9.5)	*0.56*
Plt, k/uL		75.5 (53-191)	118 (66-180)	*0.6*	65 (30-110)	43 (18-59)	*0.37*
BM blast percent, %		2.3 (0.9-6.1)	2.0 (1.0-4.3)	*0.95*	1.8 (0.3-6.7)	3.4 (1.3-9.8)	*0.24*
Therapy				*0.8*			*NA*
Supportive		15	12		2	4	
Hypomethylating agent		18	12		5	7	
Transplant		9	5		3	0	
Other		5	6		0	0	
Median follow up time, months		32.6	26.5		21.7	7.2	

		IPSS-R					
		Very low-intermediate risk			High-very high		
		No additional SNP-A abnormalities	Additional SNP-A abnormalities	*P*	No additional SNP-A abnormalities	Additional SNP-A abnormalities	*P*
Number of patients	77	26	20		18	13	
Age		67.9 (59.7-75.0)	74.0 (62.8-79.8)	*0.22*	62.5 (59.5-71.4)	69.1 (65.4-76.3)	*0.14*
Sex	77			*0.35*			*>0.99*
Male	50	15	15		12	8	
Female	27	11	5		6	5	
Initial diagnosis	77			*0.16*			*0.69*
MDS-SLD	8	5	3		0	0	
MDS-MLD	35	16	11		4	4	
MDS-EB1	12	4	1		5	2	
MDS-EB2	22	1	5		9	7	
IPSS-R		2.75 (2-3.5)	2 (1.5-3)	*0.3*	6 (5.5-6.5)	6.25 (5.6-7.9)	*0.3*
ANC, k/uL		1.6 (0.8-3.3)	2 (1.1-2.6)	*0.37*	1.2 (0.4-3.1)	0.9 (0.5-1.9)	*0.83*
Hgb, g/dL		10.1 (8.6-12.3)	10.1 (8.1-11.3)	*0.66*	8.3 (7.5-9.8)	9.0 (7.6-9.4)	*0.86*
Plt, k/uL		87.5 (69.5-164)	118 (69.8-189)	*0.62*	44 (28-81)	45 (24-122)	*0.98*
BM blast percent, %		1.6 (0.6-2.5)	1.9 (0.8-2.1)	*0.77*	8 (1.5-10)	7.4 (2.1-12)	*0.58*
Therapy				*0.99*			*0.19*
Supportive		14	10		3	6	

Hypomethylating agent	13	9	10	10
Transplant	5	3	7	2
Other	4	3	1	3
Median follow up time, months	46.2	26.5	24.2	10.3

Median value (25th-75th percentile) displayed; absolute neutrophil count (ANC), hemoglobin (Hgb), platelet count (Plt), bone marrow (BM), NA (unable to perform), "Other" therapy (therapies not listed above)

TABLE 2 Additional abnormalities detected by cytogenomic array

				SNP-A results			SNP-A and karyotype comparison		
Patient	**Karyotype**	**Cytogenetic risk**	**Cytogenetic risk after array (anomalies >5 Mb but excluding CN-LOH and small focal deletions, duplications)**	**Array results**	**%**	**Size (Mb)**	**SNP-A concordant with karyotype**	**Additional abnormality**	**Additional karyotypic abnormality not in SNP-A**
1	46,XY[20]	Good	Good	arr[hg19] 4q11q35.2(52,686,799-190,921,709)x2 hmz	90	138.2		4q CN-LOH	
2	47,XXX?c[20]	Good	Good	arr[hg19] 21q11.2q22.3(14,386,012-48,084,820)x2 hmz	30-90	33.7		21q CN-LOH	
				arr[hg19] Xp22.33q28(168,546-155,233,731)x3	100	155.1	Yes		
3	46,XY[16]	Good	Intermediate	arr[hg19] 15q14q22.2(35,437,654-61,211,671)x1-2	15	25.8		15q deletion	
4	46,XY[20]	Good	Poor	arr[hg19] 6q23.2q23.3(134,365,000-136,607,455)x1	100	2.2		6q deletion	
				arr[hg19] 7p22.3q36.3(43,360-159,119,707)x1-2	7	159.1		-7	
				arr[hg19] 11q13.1q25(65,577,515-134,942,626)x2 hmz	100	69.4		11q CN-LOH	
5	46,XX[20]	Good	Good	arr[hg19] 7q35q36.3(144,958,661-159,119,220)x2 hmz	90	14.2		7q CN-LOH	
				arr[hg19] 11q23.3(118,338,293-118,354,345)x2-3	45	16 kb		11q gain (KMT2A-PTD)	
6	46,XY[20]	Good	Good	arr[hg19] 11q12.2q25(60,804,709-134,942,626)x2 hmz	90	74.1		11q CN-LOH	
				arr[hg19] 11q23.3(118,338,293-118,349,247)x2-3	>50	11 kb		homozygous KMT2A-PTD	
7	46,XX[20]	Good	Good	arr[hg19] 11q23.3(118,123,516-118,470,527)x2-4	80	347 kb		11q gain (KMT2A-PTD)	
				arr[hg19] 11q24.3(128,408,210-128,699,707)x2-3	80	291 kb		11q24.3 gain	
8	46,XY[20]	Good	Good	arr[hg19] 1p36.33p34.1(882,802-45,000,436)x2 hmz	90	44.1		1p CN-LOH	
9	46,XY[20]	Good	Good	arr[hg19] 3q11.1q29(93,735,022-197,851,260)x2 hmz	35	104.1		3q CN-LOH	
				arr[hg19] 4q24(106,130,009-106,190,922)x1-2	50	61 kb		4q deletion including TET2	
10	46,XX[20]	Good	Good	arr[hg19] 7q21.3q36.3(97,735,123-159,119,220)x2 hmz	50	61.4		7q CN-LOH	
				arr[hg19] 21q11.2q22.3(15,867,134-48,084,820)x2 hmz	50	32.2		21q CN-LOH	
11	46,XX[20]	Good	Good	arr[hg19] 8q11.23q24.3(53,704,149-146,292,734)x2 hmz	15	92.6		8q CN-LOH	
12	46,XY[20]	Good	Good	arr[hg19] 4q22.1q35.2(92,145,040-190,921,709) x2 hmz	10	98.8		4q CN-LOH	
13	46,XY[20]	Good	Poor	arr[hg19] 7p22.3q36.3(43,360-159,119,707)x1-2	12	159.1		-7	
14	46,XY[20]	Good	Good	arr[hg19] 3q21.3q29(126,531,213-197,851,260)x2 hmz	85	71.3		3q CN-LOH	
				arr[hg19] 7q22.1(99,829,321-102,058,793)x1-2	80	2.2		7q deletion including CUX1	
				arr[hg19] 21q22.12(36,294,421-37,432,271)x1-2	85	1.1		21q deletion including RUNX1	
15	46,XX[20]	Good	Good	arr[hg19] 11q23.3(118,338,521-118,355,688)x2-3	30	17 kb		11q gain (KMT2A-PTD)	

16	46,XY[20]	Good	Good	arr[hg19] 4q24(105,995,910-106,227,999)x1-2	90	0.2		4q deletion including TET2	
17	46,XY[20]	Good	Good	arr[hg19] 21q22.12(36,002,849-37,408,933)x1-2	80	1.4		21q deletion including RUNX1	
18	46,XY[20]	Good	Good	arr[hg19] 9q34.3(139,101,277-139,734,766)x1	80	0.6		9q deletion (constitutional?)	
				arr[hg19] 11q23.3(118,335,185-118,359,052)x3	80	24 kb		11q gain (KMT2A-PTD)	
19	46,XY[20], NUP98 rearrangement (80% by FISH)	Good	Good	arr[hg19] 5q35.3(176,650,787-176,768,901)x2-3	80	0.1		5q gain	
				arr[hg19] 11p15.4(3,764,205-3,832,210)x2-3	80	68 kb		11p gain associated with NUP98-NSD1 translocation	
20	46,XY,del(5)(q15q33)[4]/46,XY[1]	Good	Good	arr[hg19] 1p36.33p31.3(903,425-65,250,982)x2 hmz	60	64.3		1p CN-LOH	
				arr[hg19] 5q15q33.2(93,274,523-154,360,732)x1-2	40	64.1	Yes		
21	46,XY,del(5)(q15q31)[19]/46,XY[1]	Good	Good	arr[hg19] 3p13p11.1(70,310,611-88,552,092)x1-2	60	18.2		3p deletion	
				arr[hg19] 5q15q31.1(95,982,582-131,916,380)x1-2	70	35.9	Yes		
22	46,XX,del(5)(q13q33)[12]/46,XX[8]	Good	Good	arr[hg19] 5q14.2q34(81,856,536-160,672,001)x1-2	50	78.8	Yes		
				arr[hg19] 12p13.31p13.1(10,000,550-13,258,017)x1-2	30	3.3		12p deletion including ETV6	
24	46,XY,del(13)(q12q14)[7]/46,XY[18]	Intermediate	Intermediate	arr[hg19] 4q24(105,942,532-106,564,759)x1-2	95	0.6		4q deletion including TET2	
				arr[hg19] 13q13.1q14.3(33,109,828-53,700,736)x1-2	25	20.6	Yes		
				arr[hg19] Xq28 or Yq12(154,941,868-155,233,731 or 59,044,874-59,336,737)x1-2	95	0.3		Xq28 or Yq12 deletion (not specific)	
26	47,XY,+8[18]/46,XY[2]	Intermediate	Intermediate	arr[hg19] 8p23.3q24.3(158,048-146,295,771)x2-3	30	146.1	Yes		
				arr[hg19] 11q13.2q25(67,015,468-134,942,626)x2 hmz	50	69.9		11q CN-LOH	
27	47,XX,+8[14]/46,XX[6]	Intermediate	Intermediate	arr[hg19] 3q26.2(168,582,060-170,230,667)x2-3	90	1.6		3q gain	
				arr[hg19] 8p23.3q11.1(158,048-47,126,524)x2-3	65	47			
				arr[hg19] 8q11.1q11.21(47,127,862-51,456,633)x1-2	90	4.3		8q deletion (+8 actually had small deletion near centromere)	
				arr[hg19] 8q11.21q24.3(51,456,754-146,295,771)x2-3	65	94.8			
29	44,XX,add(4)(q23),-5,+6,der(6;12)t(6;12)(p21;p11.2)del(6)(p12p21),der(6)t(6;13)(p21;q14)del(6)(p12p21),der(11)dup(11)(q13q13)del(11)(q23q23),-13,add(14)(p11.2),del(18)(q21q23)[cp13]/45,sl,+mar[7]	Very poor	Very poor	arr[hg19] 4q25(109,354,361-111,191,990)x1	100	1.8		4q25 deletion	
				arr[hg19] 4q32.3q35.2(167,790,246-190,957,473)x1	100	23.2	Yes		add(14p) not observed
				arr[hg19] 5p15.33p15.2(113,576-13,611,558)x1	100	13.5		monosomy 5 as 5p deletion, 5q deletion and marker	
				arr[hg19] 5q11.2q35.1(53,519,660-171,907,198)x1	100	118.4			
				arr[hg19] 6p24.1p22.1(11,797,999-27,746,178)x1	100	15.9		6p22p24 deletion	
				arr[hg19] 6p22.1p21.1(30,105,444-44,326,337)x3	100	14.2	Yes		
				arr[hg19] 6p21.1p12.1(44,326,504-56,764,165)x1	100	12.4	Yes		
				arr[hg19] 6p12.1q27(56,768,218-170,919,482)x3	100	114.2	Yes		
				arr[hg19] 11p11.12q25(51,126,723-134,938,470)cx	100	83.8		11 abnormalities complex	

				arr[hg19] 12p13.2p11.21(11,658,944-31,485,751)x1	100	19.8	Yes		
				arr[hg19] 13q11q34(19,436,286-115,107,733)cx	100	95.7		13q chromothripisis	
				arr[hg19] 17q22(54,260,365-55,515,281)x1	100	1.3		17q deletion	
				arr[hg19] 18q21.2q23(52,763,256-78,014,123)x1	100	25.3	Yes		
30	43,XY,der(3)t(3;16)(p12;q13),der(5)t(3;5)(p12;q13),-12,der(13)t(12;13)(q12;q34),der(17)t(17;20)(p13;p11.2),-20,idic(22)(p11.2)[cp18]/46,XY[2]	Very poor	Very poor	arr[hg19] 3p21.2p12.2(50,699,382-83,166,160)x1-2	65	32.5	Yes		
				arr[hg19] 3p11.2q22.3(87,417,120-136,702,414)x1-2	10	49.3		3p11.2q22.3 deletion	16q and 13q abnormalities balanced
				arr[hg19] 5q13.2q35.3(72,110,523-180,719,789)x1-2	65	108.6	Yes		
				arr[hg19] 8q24.13q24.21(126,229,398-130,825,360)x2-3	65	4.6		8q gain	
				arr[hg19] 12p13.33q12(173,786-46,058,033)x1-2	65	45.9	Yes		
				arr[hg19] 12q13.11q13.12(48,829,668-49,813,922)x1-2	65	1		12q13 deletion	
				arr[hg19] 12q21.1q21.2(75,107,892-76,608,109)x1-2	65	1.5		12q21 deletion	
				arr[hg19] 14q11.2q32.33(20,511,672-107,285,437)x2 hmz	90	86.8		14q CN-LOH	
				arr[hg19] 16p11.2(28,689,085-32,922,512)x1-2	65	4.2		16p deletion	
				arr[hg19] 17p13.3p13.1(9,474-8,172,907)x1-2	65	8.2	Yes		
				arr[hg19] 19q13.32q13.33(47,126,613-49,500,959)x1-2	65	2.4		19q deletion	
				arr[hg19] 20p12.1(15,681,353-17,022,497)x1-2	65	1.4		chromosome 20 abnormalities complex	
				arr[hg19] 20p11.21q11.21(22,735,537-29,871,042)x1-2	65	7.1			
				arr[hg19] 20q11.21(29,874,663-31,364,166)x3	100	1.5			
				arr[hg19] 20q11.21q13.33(31,382,491-62,897,159)x1-2	65	31.5			
				arr[hg19] 22q11.1q13.33(16,888,899-51,197,838)x2-3	55	34.3	Yes		
31	47,XY,t(1;3)(p13;q21),del(4)(q21q25),del(11)(q14q24),+19,del(20)(q11.2q13.3)[4]	Very poor	Very poor	arr[hg19] 4q21.23q25(84,749,459-109,483,856)x1-2	60	24.7	Yes		t(1;3) not observed
				arr[hg19] 11q14.2q24.1(86,711,530-123,487,591)x1-2	60	36.8	Yes		
				arr[hg19] 17q11.2(29,262,000-30,466,769)x1-2	60	1.2		17q deletion involving NF1	
				arr[hg19] 19p13.3q13.43(260,911-58,956,888)x2-3	20	58.7	Yes		
				arr[hg19] 20q11.21q13.31(31,062,502-56,290,652)x1-2	50	25.2	Yes		
				arr[hg19] 21q22.12(36,202,439-36,282,500)x2-3	100	80 kb		21q gain (partial duplication of RUNX1)	
				arr[hg19] Xp22.2(15,644,219-16,702,011)x1-2	50	1.1		Xp deletion involving ZRSR1	
32	44-46,XY,der(2)ins(2;6)(q23;p24p12)add(2)(q23),der(3)t(3;12)(p24;p13),add(4)(q12),der(5;22)(p10;q10),-6,der(12)t(3;12)t(?6;12)(q12;q24),der(19)dup(19)(q13	Very poor	Very poor	arr[hg19] 3p26.3p12.3(61,891-74,491,151)cx	80	74.4	Yes		
				arr[hg19] 4q12q32.2(57,131,170-163,425,170)cth	80	106.3		add(4q) chromothripisis	chromosomes 2 and 12 abnormalities to be balanced
				arr[hg19] 5q11.2q12.1(54,136,001-60,742,205)x1-2	90	6.6	Yes		

	.1q13.4)add(19)(q13.4),+mar[cp17]/43-44,sl,add(X)(p11.2),-der(2)ins(2;6)add(2),+add(2)(q32),-der(3)t(3;12),+3,+6,-7,-der(12)t(3;12)t(6;12),+add(12)(p13),add(19)(p13),add(?21)(p11.2),+del(?22)(q11.2q13),-mar[cp3]			arr[hg19] 5q12.1q35.3(61,997,673-177,260,317)x1-2	90	115.3	Yes		
				arr[hg19] 6p25.3p24.2(156,974-10,880,159)x1-2	80	10.7		6p deletion	
				arr[hg19] 16q22.3q23.2(73,602,589-80,595,329)x1-2	80	7		16q deletion	
				arr[hg19] 17p13.3p11.2(18,900-16,402,114)x2 hmz	90	16.4		17p CN-LOH	
				arr[hg19] 19q13.11q13.43(34,783,132-58,956,888)x2-3	80	24.2	Yes		
				arr[hg19] 20q11.23q13.32(34,882,014-58,360,984)x1-2	20	23.5		20q deletion	
				arr[hg19] 21q22.13q22.3(39,246,697-43,050,829)x2-3	30	3.8		21q gain	
				arr[hg19] 22q11.1q13.1(16,888,899-38,450,184)x2-3	80	21.6	Yes		
				arr[hg19] 22q13.1q13.33(39,235,339-51,197,838)x2-3	80	12	Yes		
33	44,XY,t(1;12)(q21;q24.1),-3,add(3)(p11),der(5)t(3;5)(p13;q23),-6,-7,+11,der(11)t(3;11)(q21;q13)[15]/46,XY[5]	Very poor	Very poor	arr[hg19] 1q21.3q22(153,368,019-155,017,913)x1-2	30	1.7	Yes		chromosome 11 abnormalities to be balanced
				arr[hg19] 3p26.1q29(4,103,600-197,851,936)cx	30	193.8	Yes		
				arr[hg19] 5q23.2q35.3(124,723,524-180,719,789)x1-2	30	56	Yes		
				arr[hg19] 6p25.3p22.3(156,974-24,311,197)x1-2	30	24.2	Yes		
				arr[hg19] 6p22.2p22.1(26,553,570-28,222,528)x1-2	30	1.7	Yes		
				arr[hg19] 6p21.1p12.3(43,395,395-47,462,571)x2-3	30	4.1		loss of 6 and 7 complex	+11
				arr[hg19] 6p12.3q27(47,467,934-170,919,482)x1-2	30	123.5			
				arr[hg19] 7p21.3p21.1(8,299,324-16,722,353)x1-2	30	8.4			
				arr[hg19] 7p13p12.1(44,817,005-50,784,997)x1-2	30	6			
				arr[hg19] 7q11.21(63,083,343-66,898,842)x1-2	30	3.8			
				arr[hg19] 7q35q36.3(144,075,389-159,119,707)x1-2	30	15			
				arr[hg19] 17p13.3p11.2(18,900-17,027,255)x2 hmz	40	17		17p CN-LOH	
57	46,X,del(Y)(q11.23)[8]/46,XY[12]	Good	Good	arr[hg19] Yp11.31q11.221(2,650,140-19,576,531)x1-2	40	16.9		Yp gain [del(Y) to be idic(Yq)]	
				arr[hg19] Yq11.221q11.23(19,585,828-28,799,937)x0-1	40	8.7			
69	45-46,XY,dic(3;5)(5pter->5q11.2::3p12->3q29::3p22->3pter),del(7)(q21q36),+0-1mar[cp18]/46,XY[2]	Very poor	Very poor	arr[hg19] 3p22.2p12.1(37,299,213-83,644,799)x1-2	70	46.4	Yes	5q deletion complex	
				arr[hg19] 5q11.1q12.3(49,430,268-64,262,486)x1-2	20-55	14.8	Yes		
				arr[hg19] 5q12.3q13.3(66,243,349-76,743,838)x1-2	25-70	10.5	Yes		
				arr[hg19] 5q14.3q35.3(85,913,148-180,719,789)x1-2	70	94.8	Yes		
				arr[hg19] 7q21.13q36.3(88,745,758-159,119,707)x1-2	70	70.4	Yes		
70	46~49,XX,-2,del(5)(q15q33),-6,del(6)(p23p24),-7,-13,-15,+16,del(16)(q12q24),+17,add(17)(p11.2),add(17)(p12),-18,-19,-21,+1~2r,+3~5mar[cp10]/46,XX[10]	Very poor	Very poor	arr[hg19] 2q33.1q37.3(201,635,517-242,783,384)x1-2	15	41.2	Yes		
				arr[hg19] 5q13.3q35.3(73,921,819-180,719,789)x1-2	15	106.8	Yes		
				arr[hg19] 6p22.3p22.1(18,309,604-28,848,258)x4-6	amp	10.5		6p amplification	
				arr[hg19] 13q11q34(19,436,286-115,107,733)x1-2	15	95.7	Yes		loss of 7, 15, abnormalities on

									chromosome 16 and 17 not seen by array
				arr[hg19] 19p13.2(10,856,592-12,040,283)x4-7	amp	1.2		19p amplification	
				arr[hg19] 19p13.2p13.12(13,477,560-15,274,712)x2-5	amp	1.8		19p amplification	
				arr[hg19] 21q21.1(19,629,216-20,241,815)x4-7	amp	0.6		21q amplification	
				arr[hg19] 21q22.12q22.3(35,848,786-48,097,372)x2-3	40	12.2		21q gain	
73	45,XX,der(4)t(4;?13)(q35;q14),-13,-16,der(17)t(?16;17)(p11.2;p11.2),+der(?)t(?;1)(?;p31)[18]/44-45,XX,-8,dic(8;10)(q26;q2?2)ins(10;1)(q26;p31p36),-16,der(17)t(?16;17)[cp2]	Very poor	Very poor	arr[hg19] 1p36.33p32.1(849,466-59,863,870)x2-3	80	59	Yes		the subclone -8 and dic(8;10) not observed
				arr[hg19] 1p31.1(71,990,325-73,466,675)x2-3	85	1.5	Yes		
				arr[hg19] 4q35.2(190,712,389-190,957,473)x1-2	85	0.3	Yes		
				arr[hg19] 8q23.1q24.3(108,384,827-146,292,734)x2 hmz	85	37.9		8q CN-LOH	
				arr[hg19] 13q21.31q21.33(65,565,962-69,826,807)x1-2	85	4.3		loss of 13 complex	
				arr[hg19] 13q33.1(103,508,960-103,531,681)x1-2	85	23 kb			
				arr[hg19] 13q33.1(103,592,015-104,256,190)x1-2	85	0.7			
				arr[hg19] 13q33.2q33.3(105,264,673-109,888,382)x1-2	85	4.6			
				arr[hg19] 16q11.2q24.3(46,503,572-90,155,062)x1	100	43.7	Yes		
				arr[hg19] 17p13.3p11.2(525-17,988,254)x1	95	18	Yes		
74	45,XY,del(5)(q22q33),der(17;20)(q10;p10)[3]/46,sl,del(7)(q11.2q36),+8[5]/44,sl,-7[6]/85-94,slx2,add(11)(q12)x2,+1-2mar,4-11dmin[cp4]/46,XY[2]	Very poor	Very poor	arr[hg19] 5q21.3q33.3(106,203,054-157,533,436)x1-2	30	51.3	Yes		
				arr[hg19] 7p22.3q36.3(43,360-159,119,707)x1-2	15	159.1	Yes		
				arr[hg19] 8p23.3q24.3(158,048-146,295,771)x2-3	10	146	Yes		
				arr[hg19] 11q13.4(70,735,084-74,545,922)x1-2	10	3.8			
				arr[hg19] 11q13.4q14.1(74,950,287-80,597,434)x2-3	10	5.7		add(11q) complex	near tetraploid clone not observed
				arr[hg19] 11q14.1q14.3(80,885,537-92,686,784)x1-2	10	11.8			
				arr[hg19] 11q22.1q22.3(97,942,172-103,313,943)x1-2	10	5.4			
				arr[hg19] 11q22.3q23.3(104,772,138-117,129,522)x1-2	10	12.4			
				arr[hg19] 11q23.3(117,940,196-118,570,397)x2-3	80	0.6			
				arr[hg19] 11q23.3q24.2(118,719,566-126,066,194)x1-2	10	7.4			
				arr[hg19] 11q24.2q25(126,201,307-134,938,470)x3-5	amp	8.7			
				arr[hg19] 17p13.3p11.2(525-21,722,139)x1-2	30	21.7	Yes		
				arr[hg19] 20q11.22q13.33(32,359,017-62,915,555)x1-2	30	32.6	Yes		
				arr[hg19] 21q21.3q22.3(30,253,288-48,097,372)x2-3	10	17.8		21q gain	
76	46-48,XX,del(1)(p22p36.1),del(2)(p24),del(3)(p21),-	Very poor	Very poor	arr[hg19] 1p21.1p13.3(103,620,493-108,243,851)x1-2	70	4.6	Yes		
				arr[hg19] 3q26.33(180,626,349-180,728,459)x1-2	70	0.1		3q deletion	

	5,der(6)t(1;6)(p13;q25),der(7)add(7)(p21)del(7)(q32q36),+8,+8,+8,del(11)(q22q23),-12,+13,del(13)(q12q14)x2,add(16)(q11.2),add(17)(p13),add(19)(p13),-21, +0-2mar[cp20]			arr[hg19] 5p15.33p14.1(113,576-24,705,254)x1-2	80	24.6		loss of 5 complex	
				arr[hg19] 5p14.1p13.2(26,602,764-36,629,258)x1-2	80	10			
				arr[hg19] 5q14.3(86,075,402-91,536,195)x1-2	80	5.5			
				arr[hg19] 5q21.1q32(99,149,791-149,316,740)x1-2	80	50.2			
				arr[hg19] 5q33.1q34(151,060,202-163,263,177)x1-2	80	12.2			
				arr[hg19] 6q11.1q13(62,118,669-73,010,280)x1-2	80	10.9	Yes		
				arr[hg19] 6q24.2q25.1(144,387,634-152,252,413)x1-2	80	7.9	Yes		
				arr[hg19] 7p21.3p21.1(11,371,004-17,035,173)x1-2	80	5.7	Yes		
				arr[hg19] 7p14.1(37,771,693-40,371,298)x1-2	80	2.6	Yes		
				arr[hg19] 7q33q36.3(134,801,961-159,119,707)x1-2	80	24.3	Yes		
				arr[hg19] 8p23.3q24.3(158,048-146,295,771)x3-4	pentasomy	146.1	Yes		
				arr[hg19] 12p12.3q22(15,277,112-95,085,949)cx	80	79.8		loss of 12 complex	
				arr[hg19] 13q11q13.1(19,436,286-32,565,539)x2-3	70	13.1	Yes		
				arr[hg19] 13q13.1q21.31(32,576,910-64,735,038)x1-2	70	32.2	Yes		
				arr[hg19] 13q21.31q34(64,738,525-115,107,733)x2-3	70	50.4	Yes		
				arr[hg19] 16q12.1(48,341,743-50,857,833)x1-2	80	0.4		add(16q) complex	
				arr[hg19] 16q12.1q24.3(51,570,940-90,155,062)x1-2	80	2.5			
				arr[hg19] 16q11.2q12.1(46,580,413-47,022,778)x1-2	80	38.6			
				arr[hg19] 17p13.3p11.2(18,900-20,697,797)x2 hmz	80	20.7		17p CN-LOH	loss of 21 not observed

Copy neutral loss of heterozygosity (CN-LOH), partial tandem duplication (PTD), amplification (amp)