BOBS ASSAY FOR PRENATAL DIAGNOSIS OF SOME ANEUPLOIDIES AND MICRODELETIONS

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OVERVIEW

- Prenatal diagnosis of genetic dissoder is necessary.

- Today, cytogenetic and QF-PCR tests are common in prenatal diagnosis, but it can’t detect chromosomal microdeletions.

- BoBs (Bacs-on-Beads) assay can detect common aneuploidies *13*, *18*, *21, X, Y* chromosomes and other nine microdeletions for rapid prenatal diagnosis.

Aims

- *Detection of some aneuploidies and microdeletions in prenatal diagnosis by BoBs assay.*
- *Assess prenatal diagnosis results by BoBs assay.*

SAMPLES AND METHODS

- Samples: 30 amniotic fluid samples from single pregnant women in ≥ 16 week of gestation with abnormal fetal ultrasounds.

- Methods: cross-sectional study. Each Sample was tested by both BoBs and cytogenetic technique.

Results and Disscusion

3.1. Chromosomal dissorders were deteted by BoBs and cytogenetics

Table 3.1. **Comparing results of BoBs and results of cytogenetics**

Karyotype	BoBs		Karyotype	
	n	Ratio (%)	n	Ratio(%)
46,XX (XY)	24	80	27	90
Trisomy 18	1	3,33	1	3,33
Trisomy 21	2	6,67	2	6,67
Microdeletions	3	10	0	0
Total	30	100	30	100

Results and Disscusion

3.2. Comparing results of genetic abnormalities and prenatal screening test

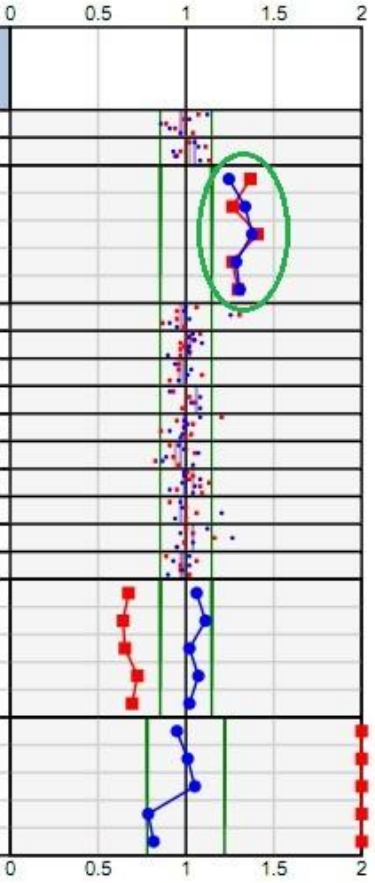
Table 3.2. **Comparing results of cytogenetics, BoBs asay and prenatal screening test**

TT	Ultrasound	Test of maternal serum screen	BoBs	Karyotype
1	Ventricular septal defect	NI	47,XX, + 21	47,XX, + 21
2	Ventricular septal defect, Polyhydramnios	NI	47,XX, + 18	47,XX, + 18
3	Tetralogy of Fallot	NI	Digeogre	46,XY
4	Ventricular septal defect	NI	Digeogre	46,XY
5	Increase NT Ventricular septal defect	NI	47,XY, + 21	47,XY, + 21
6	Neural tube defect	High risk of Down syndrome 1/26	Cri-du Chat	46,XX

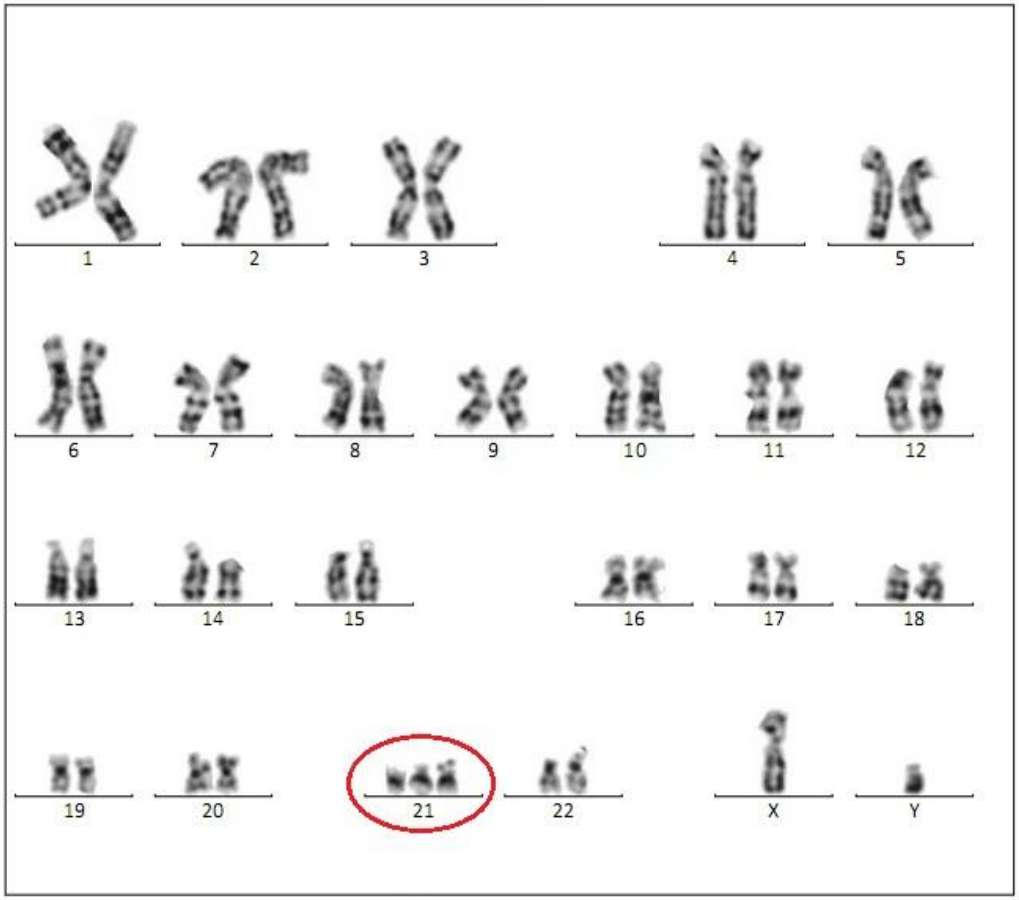
Result BoBs : Trisomy 21

Normalized Ratios

Probe	/F	/M	Chromosomal Location (NCBI37.3)
13C	0.99	0.97	
18C	1.02	1.05	
21C1; 74 21q22	1.37	1.24	32.208- 32.373
21C2; 75 21q22	1.26	1.34	34.661- 34.820
21C3; 76 21q22	1.41	1.38	35.229- 35.372
21C4; 77 21q22	1.26	1.29	43.556- 43.770
21C5; 78 21q22	1.30	1.31	44.146- 44.311
AUTO	0.97	0.99	
CDC	1.00	1.02	
DGS	0.96	0.98	
DiG	1.00	1.06	
LGS	1.00	0.99	
MDS	0.94	0.97	
PWS	1.04	0.99	
SMS	1.00	0.97	
WBS	1.00	1.04	
WHS	0.97	0.98	
XC1; 79 Xp22	0.67	1.06	6.958- 7.137
XC2; 80 Xp22	0.64	1.11	10.699- 10.763
XC3; 81 Xp21	0.65	1.02	37.368- 37.555
XC4; 84 Xq13	0.72	1.07	73.303- 73.450
XC5; 85 Xq27	0.69	1.02	146.757- 146.960
YC1; 86 Yp11	5.07	0.95	8.461- 8.636
YC2; 96 Yq11	5.92	1.01	15.036- 15.174
YC3; 87 Yq11	10.61	1.05	19.299- 19.472
YC4; 88 Yq11	5.24	0.79	25.313- 25.416
YC5; 89 Yq11	4.50	0.82	25.489- 25.646



Karyotype: Trisomy 21

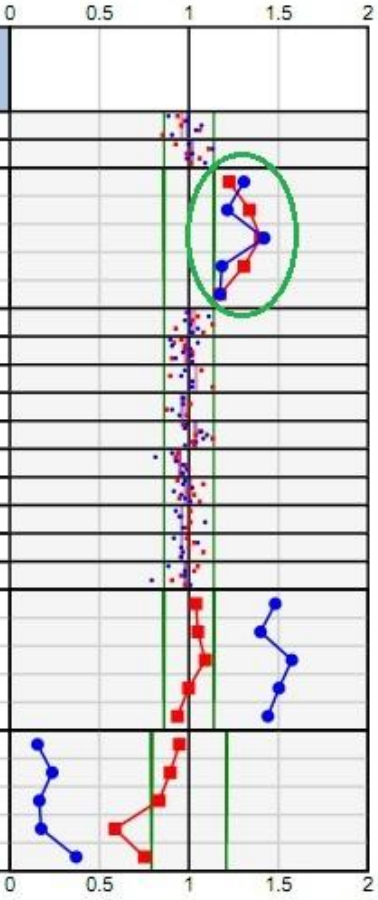


KARYOTYPE: 47,XY,+21

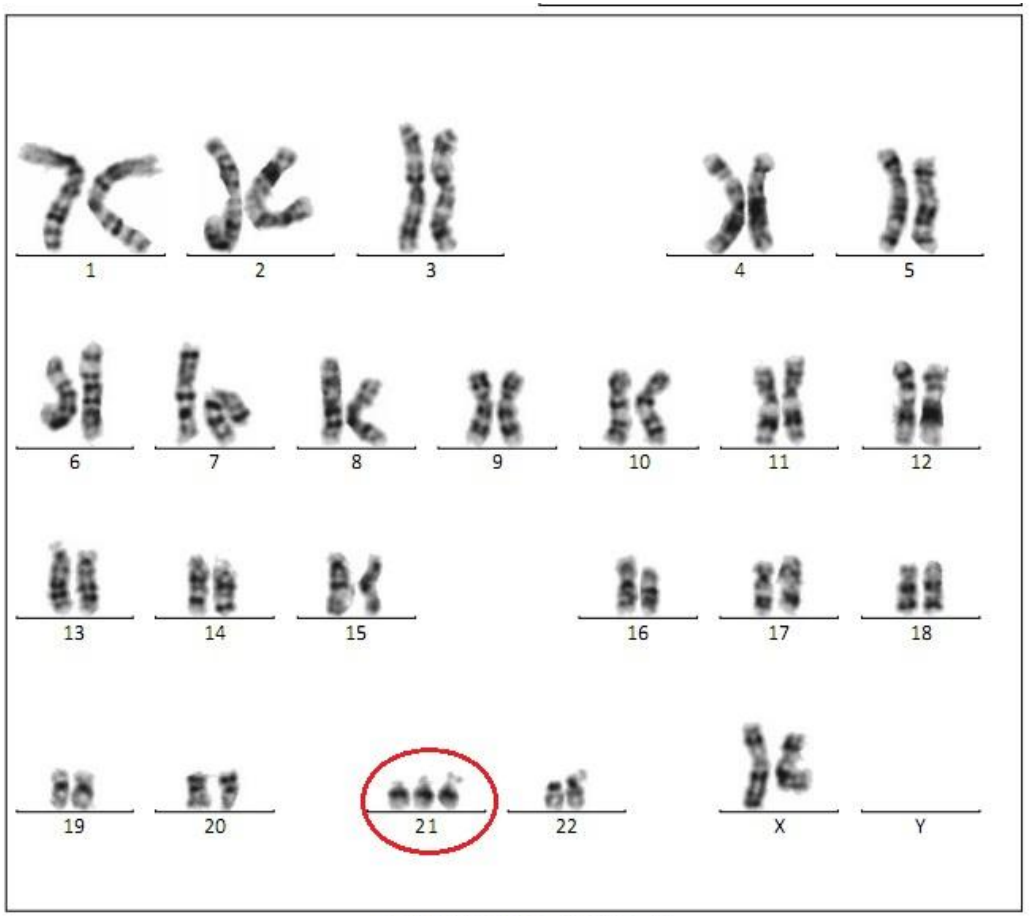
Result BoB: trisomy 21

Normalized Ratios

Probe	/F	/M	Chromosomal Location (NCBI37.3)
13C	0.96	0.99	
18C	1.01	1.02	
21C1; 74 21q22	1.22	1.31	32.208- 32.373
21C2; 75 21q22	1.34	1.21	34.661- 34.820
21C3; 76 21q22	1.40	1.42	35.229- 35.372
21C4; 77 21q22	1.31	1.18	43.556- 43.770
21C5; 78 21q22	1.17	1.17	44.146- 44.311
AUTO	1.03	1.01	
CDC	0.98	0.99	
DGS	1.04	1.00	
DiG	0.98	0.96	
LGS	1.03	1.04	
MDS	0.95	0.95	
PWS	1.01	0.98	
SMS	1.00	0.96	
WBS	1.00	0.96	
WHS	0.98	0.98	
XC1; 79 Xp22	1.04	1.48	6.958- 7.137
XC2; 80 Xp22	1.05	1.40	10.699- 10.763
XC3; 81 Xp21	1.09	1.57	37.368- 37.555
XC4; 84 Xq13	1.00	1.50	73.303- 73.450
XC5; 85 Xq27	0.94	1.44	146.757- 146.960
YC1; 86 Yp11	0.95	0.15	8.461- 8.636
YC2; 96 Yq11	0.90	0.24	15.036- 15.174
YC3; 87 Yq11	0.83	0.16	19.299- 19.472
YC4; 88 Yq11	0.59	0.17	25.313- 25.416
YC5; 89 Yq11	0.75	0.37	25.489- 25.646



Karyotype: Trisomy 21

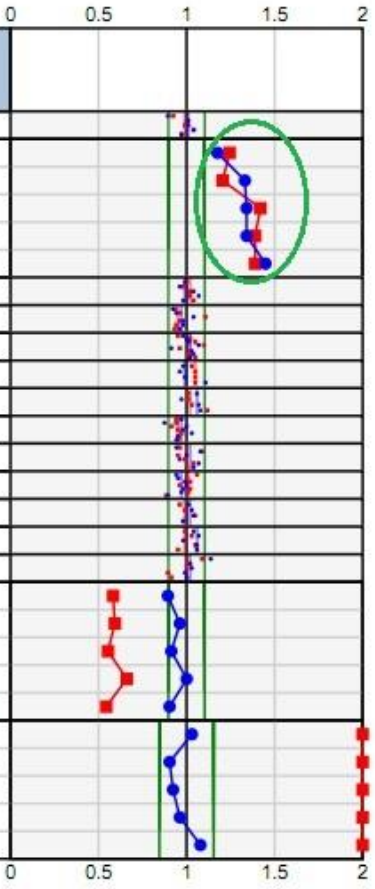


KARYOTYPE: 47,XX,+21

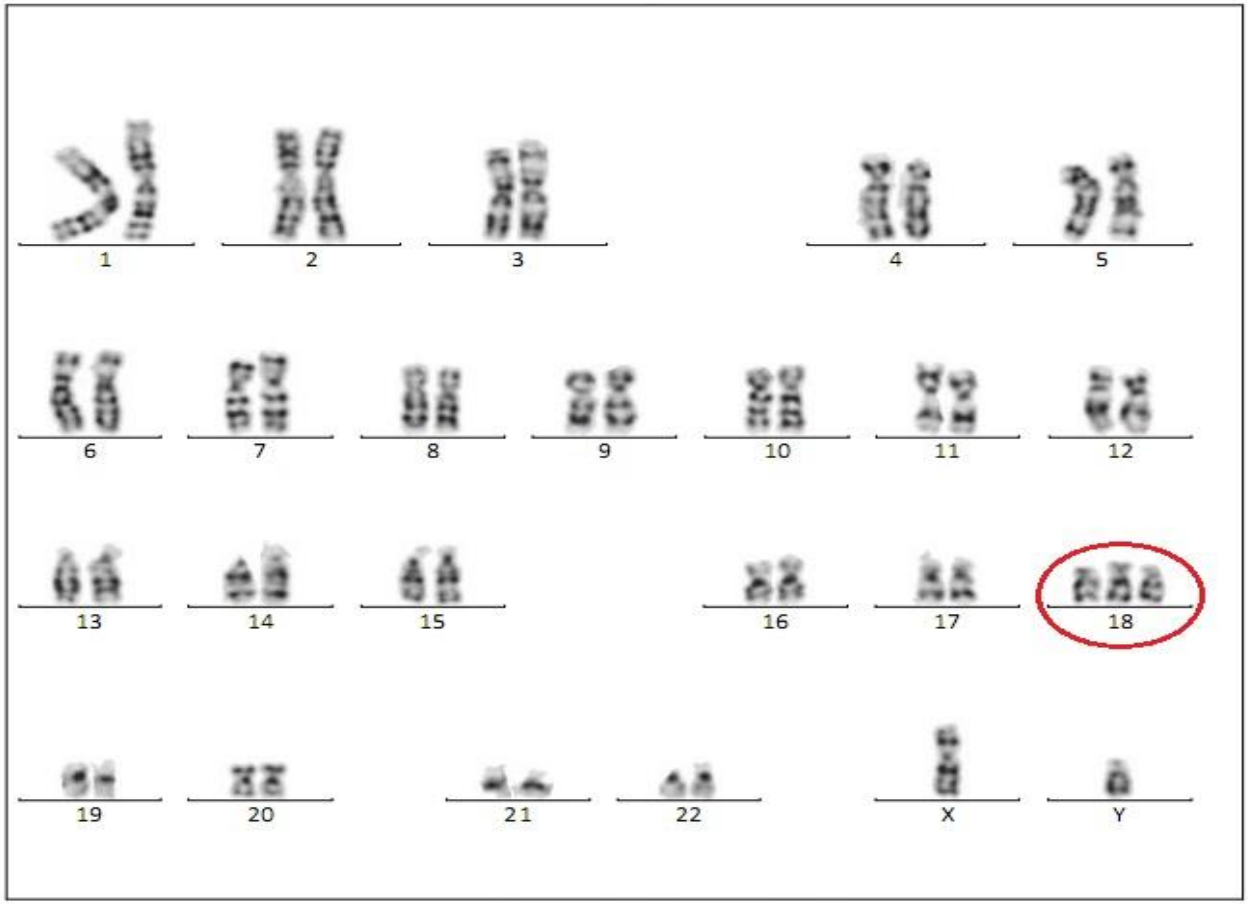
Result of BoBs: Trisomy 18

Normalized Ratios

Probe	/F	/M	Chromosomal Location (NCBI37.3)
13C	0.99	1.01	
18C1; 67 18p11	1.24	1.18	2.735- 2.908
18C2; 68 18p11	1.21	1.33	13.989- 14.143
18C3; 69 18q12	1.42	1.34	30.827- 30.986
18C4; 70 18q21	1.39	1.34	51.861- 52.034
18C5; 73 18q22	1.39	1.45	61.790- 61.971
21C	1.02	0.99	
AUTO	0.95	0.97	
CDC	1.02	1.02	
DGS	1.05	0.99	
DiG	1.02	1.06	
LGS	0.95	0.97	
MDS	1.00	1.02	
PWS	1.01	0.97	
SMS	0.99	1.02	
WBS	0.99	1.06	
WHS	1.00	1.02	
XC1; 79 Xp22	0.58	0.89	6.958- 7.137
XC2; 80 Xp22	0.59	0.96	10.699- 10.763
XC3; 81 Xp21	0.55	0.91	37.368- 37.555
XC4; 84 Xq13	0.66	1.00	73.303- 73.450
XC5; 85 Xq27	0.54	0.90	146.757- 146.960
YC1; 86 Yp11	7.89	1.03	8.461- 8.636
YC2; 96 Yq11	5.51	0.90	15.036- 15.174
YC3; 87 Yq11	11.58	0.92	19.299- 19.472
YC4; 88 Yq11	6.22	0.96	25.313- 25.416
YC5; 89 Yq11	4.84	1.08	25.489- 25.646



Karyotype : Trisomy 18

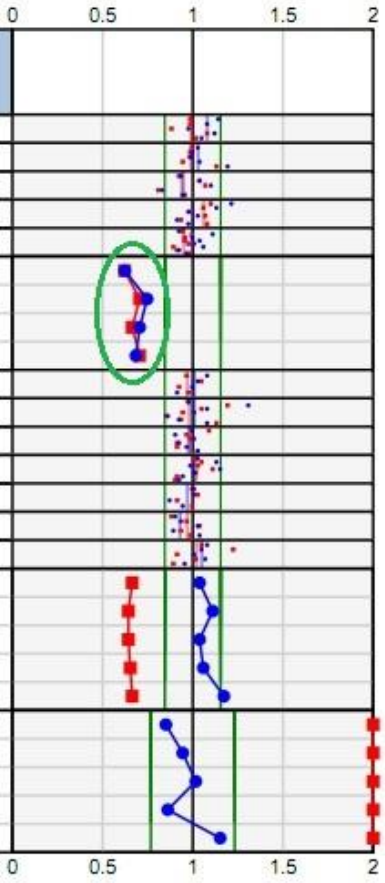


KARYOTYPE: 47,XY,+18

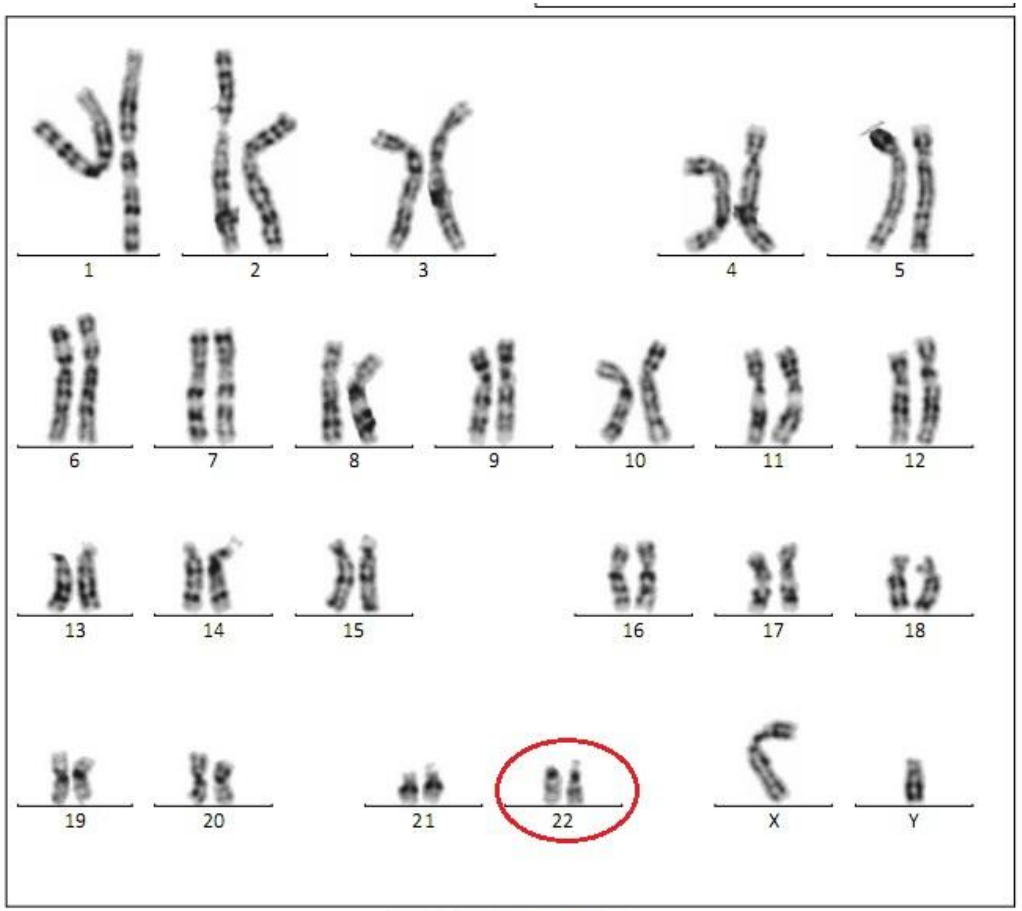
Result of BoBs : Digeogre

Normalized Ratios

Probe	/F	/M	Chromosomal Location (NCBI37.3)
13C	0.99	1.08	
18C	0.99	1.03	
21C	0.95	0.94	
AUTO	1.06	1.00	
CDC	0.95	1.00	
DGS1; 16 22q11	0.62	0.62	17.736- 17.777
DGS2; 17 22q11	0.71	0.75	17.865- 18.040
DGS3; 18 22q11	0.66	0.71	18.073- 18.287
DGS4; 19 22q11	0.71	0.68	18.138- 18.300
DiG	0.97	1.01	
LGS	0.98	1.01	
MDS	0.98	0.98	
PWS	1.02	1.01	
SMS	1.00	0.97	
WBS	0.97	0.93	
WHS	1.02	1.05	
XC1; 79 Xp22	0.66	1.04	6.958- 7.137
XC2; 80 Xp22	0.64	1.11	10.699- 10.763
XC3; 81 Xp21	0.64	1.04	37.368- 37.555
XC4; 84 Xq13	0.65	1.06	73.303- 73.450
XC5; 85 Xq27	0.66	1.17	146.757- 146.960
YC1; 86 Yp11	7.06	0.85	8.461- 8.636
YC2; 96 Yq11	6.11	0.94	15.036- 15.174
YC3; 87 Yq11	9.13	1.02	19.299- 19.472
YC4; 88 Yq11	4.30	0.86	25.313- 25.416
YC5; 89 Yq11	5.43	1.15	25.489- 25.646



Karyotype: Digeogre

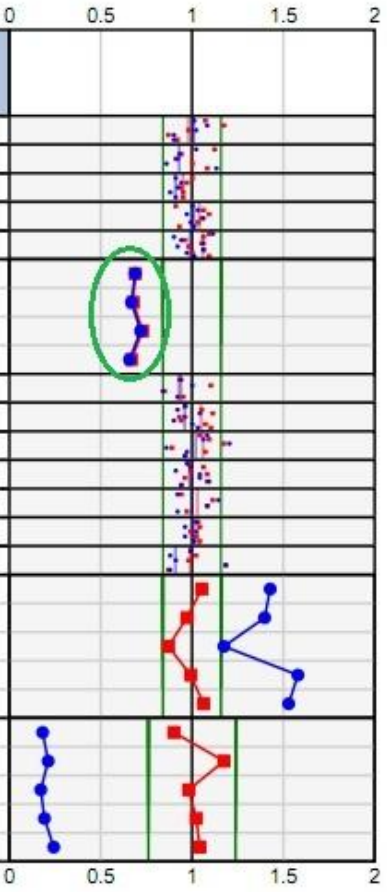


KARYOTYPE: 46,XY

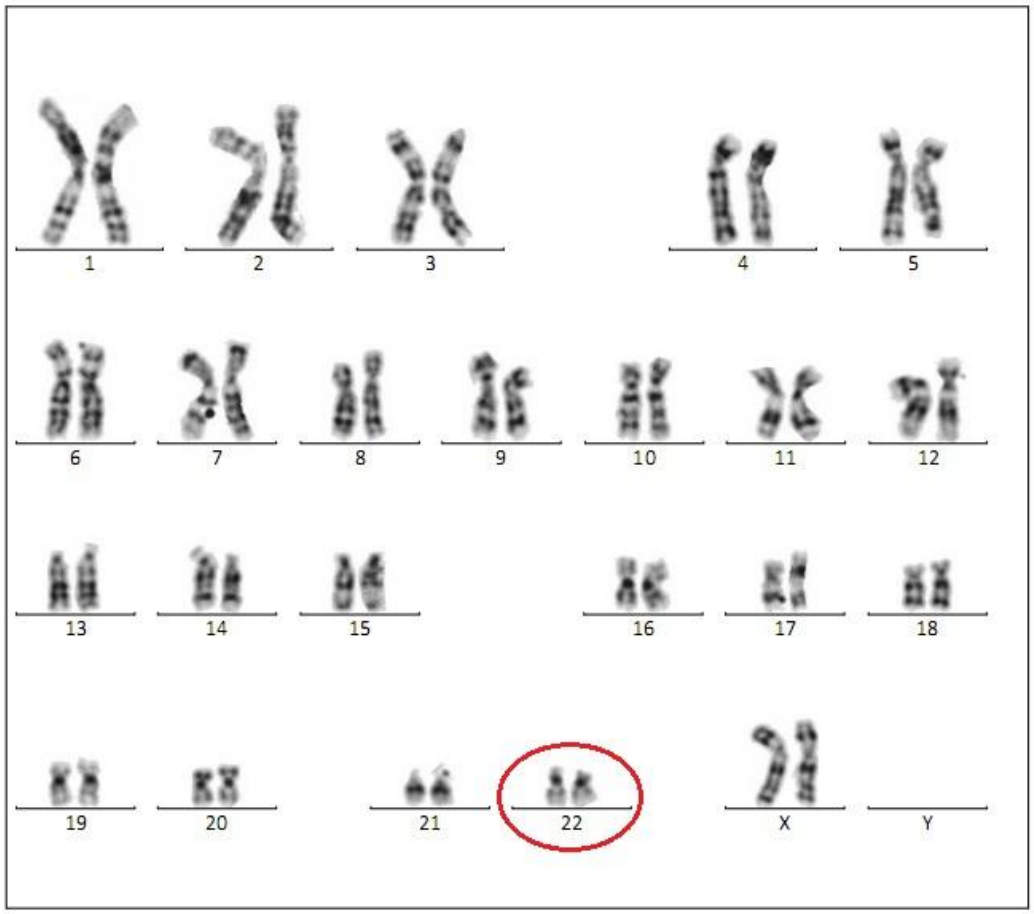
Result of BoBs : Digeogre

Normalized Ratios

Probe	/F	/M	Chromosomal Location (NCBI37.3)
13C	0.98	1.01	
18C	1.00	0.93	
21C	0.95	0.91	
AUTO	1.04	1.01	
CDC	1.06	0.99	
DGS1; 16 22q11	0.69	0.69	17.736- 17.777
DGS2; 17 22q11	0.68	0.67	17.865- 18.040
DGS3; 18 22q11	0.73	0.72	18.073- 18.287
DGS4; 19 22q11	0.67	0.66	18.138- 18.300
DiG	0.94	0.93	
LGS	1.05	0.96	
MDS	1.06	1.02	
PWS	0.99	0.99	
SMS	1.03	1.00	
WBS	1.03	1.01	
WHS	0.99	0.91	
XC1; 79 Xp22	1.05	1.43	6.958- 7.137
XC2; 80 Xp22	0.97	1.40	10.699- 10.763
XC3; 81 Xp21	0.87	1.17	37.368- 37.555
XC4; 84 Xq13	0.99	1.58	73.303- 73.450
XC5; 85 Xq27	1.06	1.53	146.757- 146.960
YC1; 86 Yp11	0.90	0.18	8.461- 8.636
YC2; 96 Yq11	1.17	0.21	15.036- 15.174
YC3; 87 Yq11	0.98	0.17	19.299- 19.472
YC4; 88 Yq11	1.02	0.19	25.313- 25.416
YC5; 89 Yq11	1.04	0.24	25.489- 25.646



Karyotype Digeogre

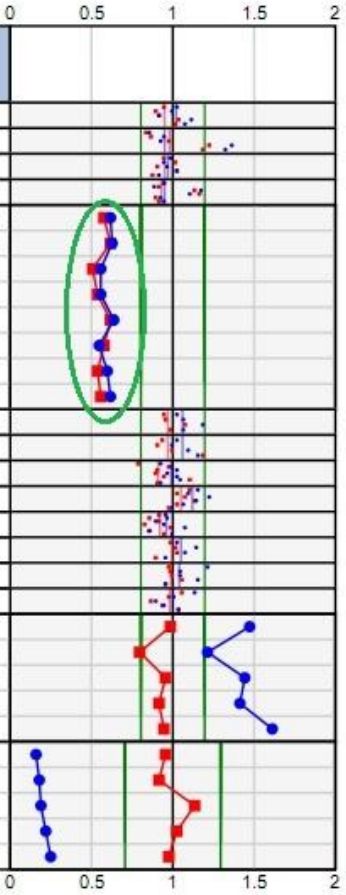


KARYOTYPE: 46,XX

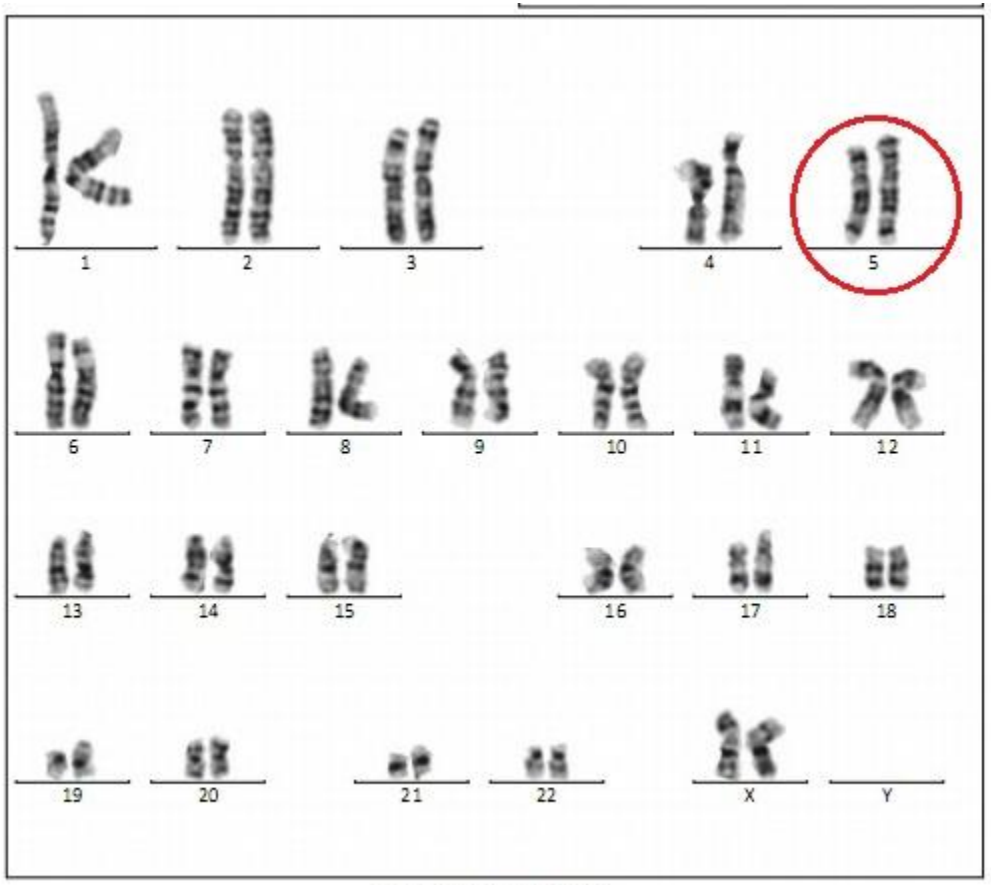
Result of BoBs : Cri-du Chat

Normalized Ratios

Probe	/F	/M	Chromosomal Location (NCBI37.3)
13C	0.95	1.02	
18C	0.98	0.95	
21C	0.95	0.97	
AUTO	0.93	0.95	
CDC1; 8 5p15	0.58	0.62	3.055- 3.214
CDC2; 9 5p15	0.62	0.63	4.545- 4.707
CDC3; 10 5p15	0.51	0.56	5.879- 6.020
CDC4; 11 5p15	0.54	0.56	6.357- 6.533
CDC5; 12 5p15	0.62	0.64	8.165- 8.312
CDC6; 13 5p15	0.58	0.55	9.086- 9.240
CDC7; 14 5p15	0.54	0.60	9.866- 10.044
CDC8; 15 5p15	0.56	0.62	10.622- 10.774
DGS	0.97	1.06	
DiG	0.97	1.06	
LGS	0.91	0.99	
MDS	1.05	1.12	
PWS	0.92	0.96	
SMS	1.00	1.05	
WBS	0.99	1.04	
WHS	0.98	1.00	
XC1; 79 Xp22	0.99	1.47	6.958- 7.137
XC2; 80 Xp22	0.80	1.21	10.699- 10.763
XC3; 81 Xp21	0.96	1.44	37.368- 37.555
XC4; 84 Xq13	0.92	1.41	73.303- 73.450
XC5; 85 Xq27	0.95	1.61	146.757- 146.960
YC1; 86 Yp11	0.96	0.16	8.461- 8.636
YC2; 96 Yq11	0.92	0.18	15.036- 15.174
YC3; 87 Yq11	1.13	0.19	19.299- 19.472
YC4; 88 Yq11	1.02	0.22	25.313- 25.416
YC5; 89 Yq11	0.98	0.25	25.489- 25.646



Karyotype Cri-du Chat



KARYOTYPE: 46,XX

Conclusions

- Three of 30 samples were detected chromosomal dissorders by cytogenetic assay (2 Trisomy 21; 1 trisomy 18)

- Six of 30 samples were detected chromosomal dissorders by BOBS assay (2 Trisomy 21; 1 trisomy 18 and 3 microdeletions).

Thank you for attention!