Preimplantasyon GenetikTanı

Tek Gen Hastalıkları ve HLA Haplotiplemesi

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PGT endikasyonları

- Otozomal Resesif ve Dominant Hastalıklar
- X-link Hastalıklar
- Kanser Predispozisyon Genleri
- Maternal-Fetal İnkompatibilite
- HLA genotiplemesi
- Tek Gen Hastalığı ve Anöploidi Birlikte Testi

Anöploidi Testi : İleriAnne Yaşı
Tekrarlayan IVF Başarısızlıkları
Tekrarlayan Düşükler

Kromozomal Yeni Düzenlenimler

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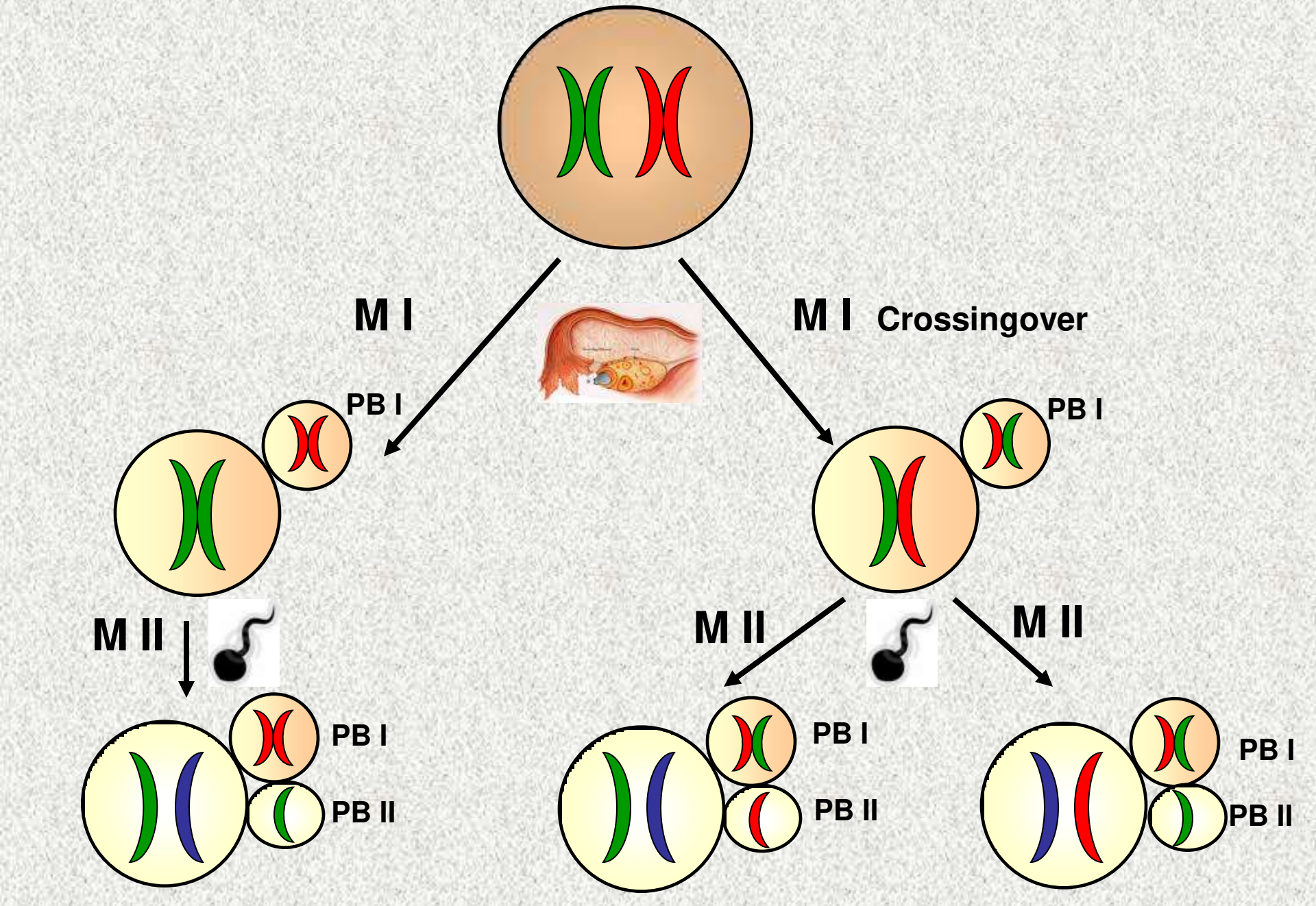
MAYOZ I ve II


M I
M I Crossingover
PB I
PB I
M II
M II
M II
PB I
PB II
PB I
PB II
PB I
PB II

PGT Klinik Uygulama Şeması

Gün 0 | Gün 1 | Gün 3 | Gün 3-6

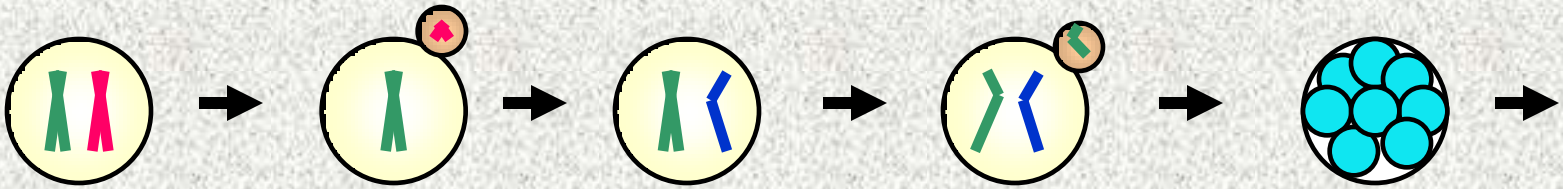
Oosit Toplanması

Polar Body I Biopsisi

Sperm Enjeksiyonu (ICSI)

Polar Body II Biopsisi

Blastomer Biopsisi



Embriyo Transferi

PCR FISH

PCR FISH

PB I + II Analizi

Blastomer Analizi

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PGT Tek Gen Hastalıkları Stratejileri

PCR

FISH

MATERNAL Otozomal Dominant (MD, PKD1, SCA, TS)	**PB + BB**	**BB**
PATERNAL Otozomal Dominant (CMT1A, APC, NF1)	**BB**	**PB**
Otosomal Resesif (CF, β-Thal, SMA)	**PB + BB**	**BB**
X-link (DMD, Frajil-X, ALD)	**PB + BB**	**BB**
HLA	**PB + BB**	
Anöploidi	**BB**	

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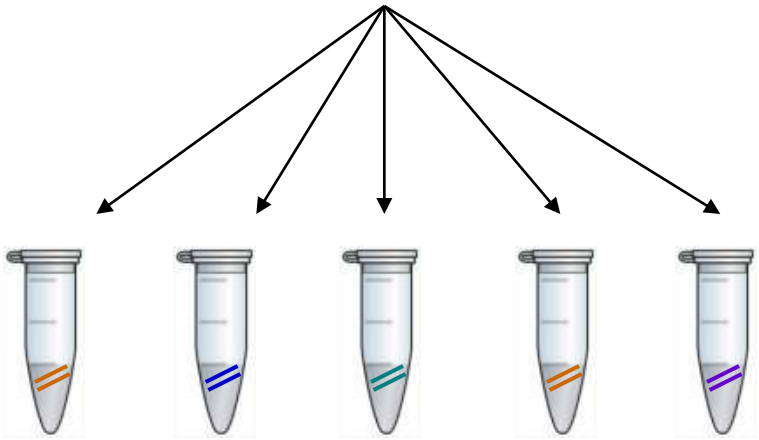
Tek-Hücre Multipleks Nested PCR

Tek Hücre (PB or Blastomer)

Hücre Parçalanması (Proteinaz K ile)

1nci Round Multipleks PCR solüsyonu (Aynı tüpte dış primerlerin hepsini içerir)

2nci Round PCR (Ayrı tüplerde iç primerleri içerir)



Gen Mutasyonu **Link Genetik Markerler**

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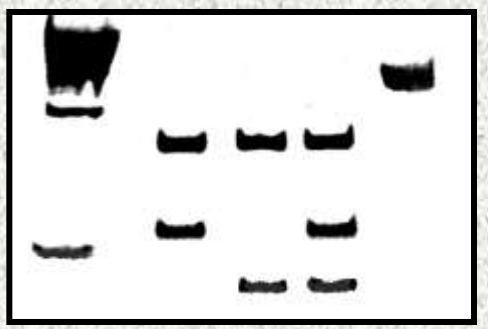
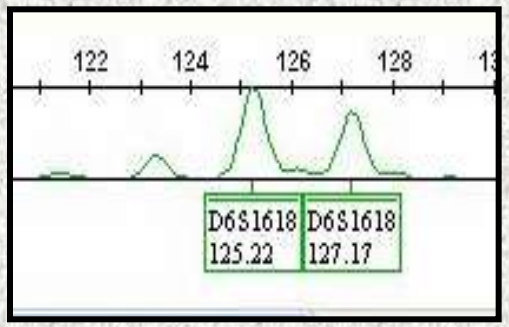
Detection of PCR products

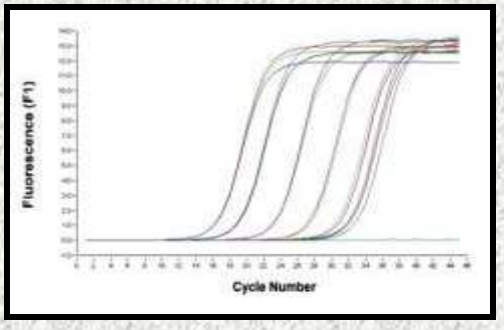
RFLP



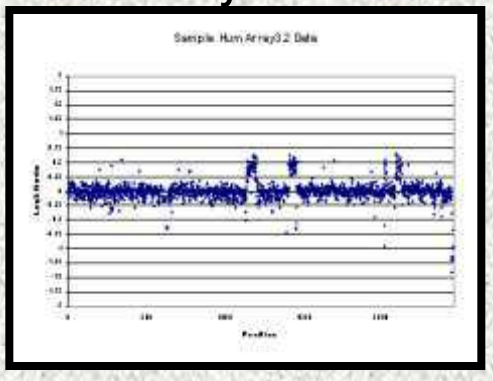
Floresan Genotipleme



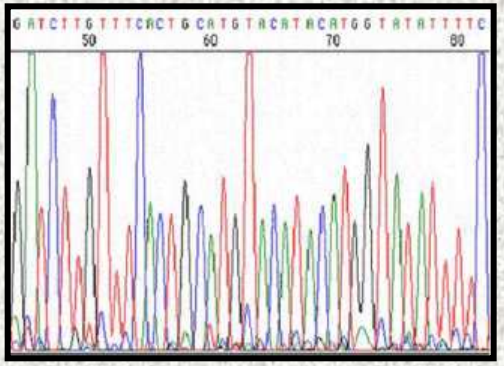
Real Time PCR



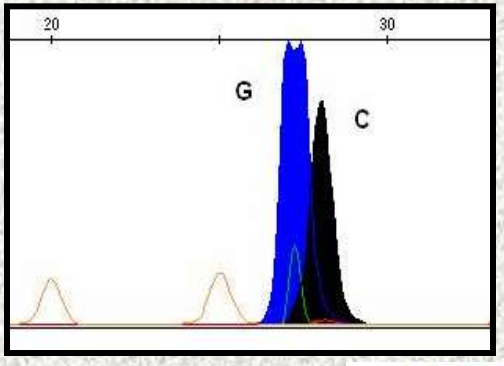
Array CGH



DNA Dizi Analizi



Minisekanslama



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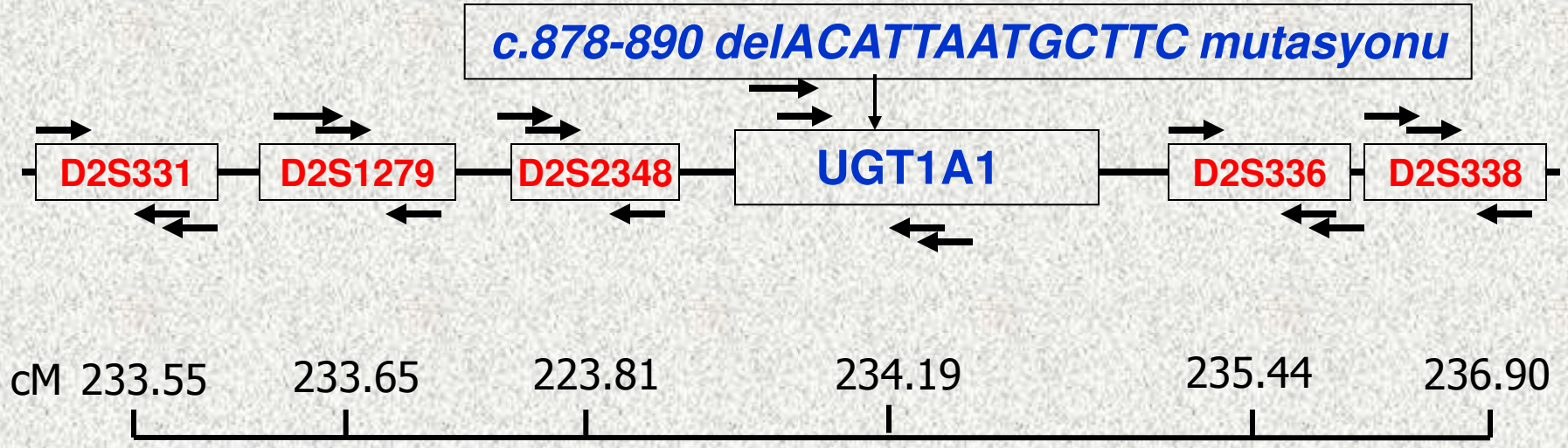
PGT Protokol Hazırlanması

2q37 bölgesi Genomik Organizasyonu (Crigler-Najjar Sendromu, UGT1A1 lokusü)



cM mesafesi her genetik markerin 2 Nolu Kromozom üzerindeki yerini belirtmektedir

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PGT Protokol Optimizasyonu

UGT1A1 gen, c.878-890 delACATTAATGCTTC mutasyonu

Primer -1 ***Primer -3***

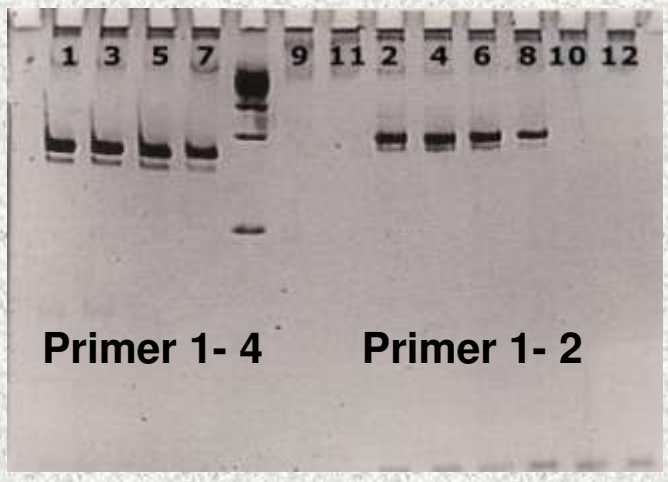
TGGTTTTTGTTGGTGGAATCAACTGCCTTCACCAAAATCCACTATCCCAGGAATTTGAAGCCTACATTAATGCTTCTGGAGAACATGGAATTGTGGTTTTCTCTTTGGGATCAATGGTCTCAGAAATTCCAGAGAAGAAAGCTATGGCA

Primer -4 **Primer -2**

Gradiyent PCR

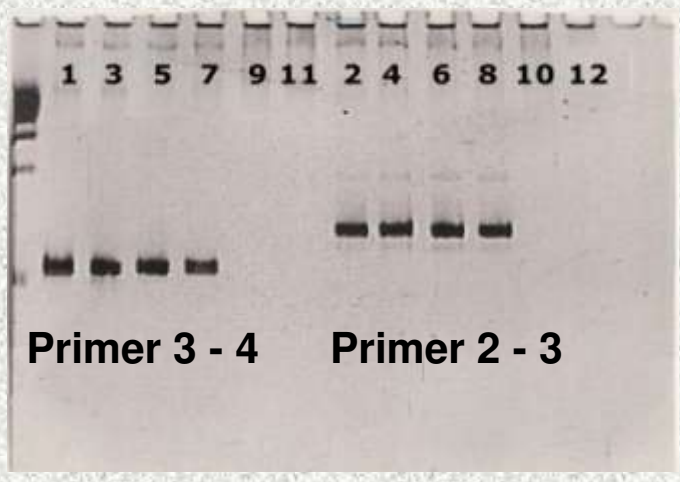
Annealing t °C

47.4 48.4 51.4 55.3 59.6 61.7 47.8 49.7 53.4 57.3 61.0 62.2



Annealing t °C

47.4 48.4 51.4 55.3 59.6 61.7 47.8 49.7 53.4 57.3 61.0 62.2



En iyi PCR Primer Kombinasyonları ve Optimal Tm °C

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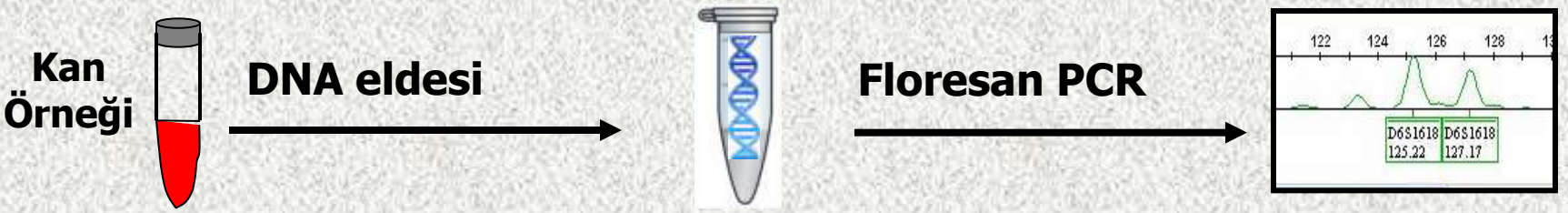
Aile Bireylerinde Haplotipleme Yapılması

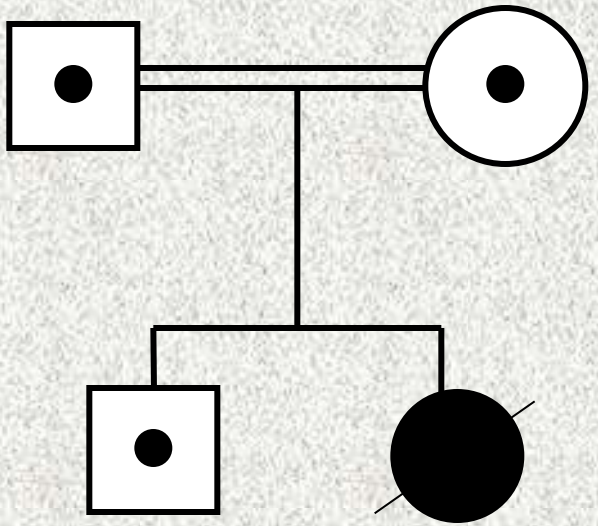


Paternal Haplotip

134	140
113	101
174	176
DEL	WT
196	208
112	116

Maternal Haplotip

111	134
115	113
174	174
WT	DEL
184	196
112	112



Genetik Markerlerin Sırası

1. D2S331
2. D2S1279
3. D2S2348
4. UGT1A1
5. D2S336
6. D2S338

140	134
101	113
176	174
WT	DEL
208	196
116	112

134	134
113	113
174	174
DEL	DEL
196	196
112	112

→ Maternal Affekte Kromozom

→ Paternal Affekte Kromozom

→ Maternal Normal Kromozom

→ Paternal Normal Kromozom

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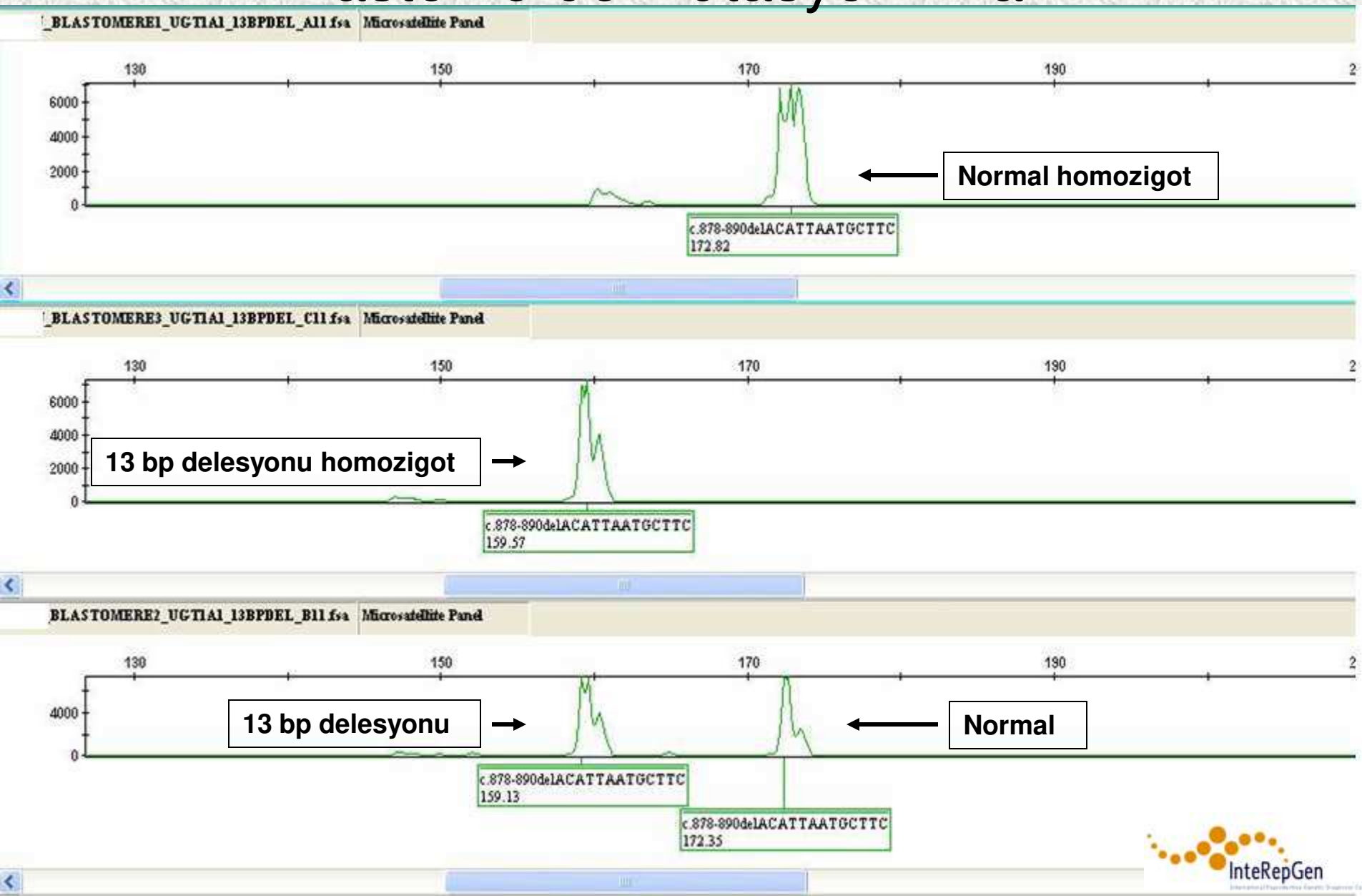
PGT Vaka Performansı
Blastomerde Mutasyon Analizi



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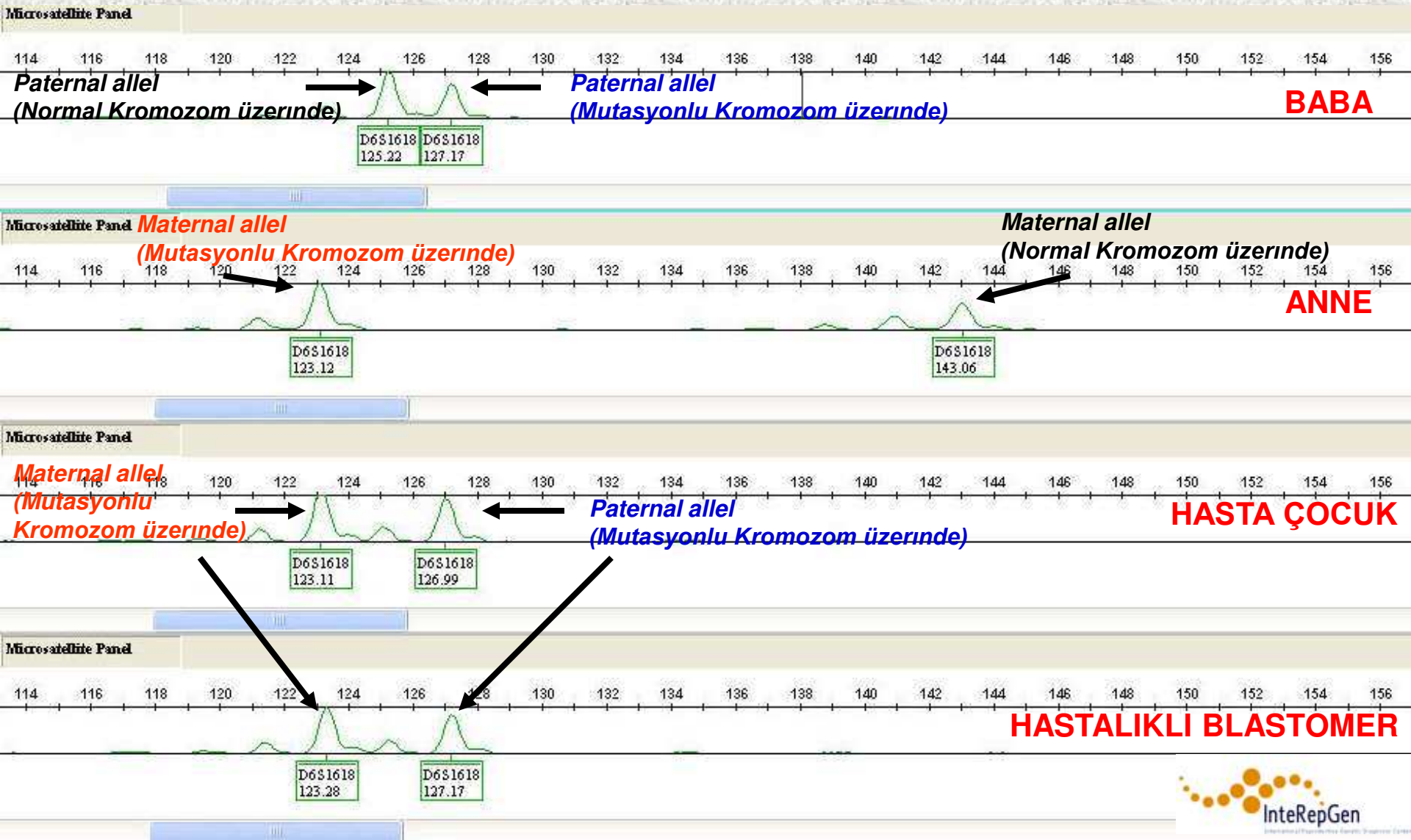
TEK SPERM HAPLOTİPLEME SONUÇLARI

SPERM #	UGT1A1 877 T/A	UGT1A1 878-890 del ACATTAATGCTTC	D2S331	D2S1279	D2S2348	D2S336	D2S338	GENOTIP
1	-	-	-	-	-	-	-	Sonuç Yok
2	A	878-890 del ACATTAATGCTTC	148	155	187	189	99	MUTASYONLU
3	-	-	-	-	-	-	-	Sonuç Yok
4	A	878-890 del ACATTAATGCTTC	148	155	187	185	97	MUTASYONLU ve Rekombinant
5	T	NORMAL	ADO	188	ADO	185	97	NORMAL
6	-	-	-	-	-	-	-	Sonuç Yok
7	-	-	-	-	-	-	-	Sonuç Yok
8	A	878-890 del ACATTAATGCTTC	148	155	187	189	99	MUTASYONLU
9	T	NORMAL	139	188	183	185	97	NORMAL
10	T	NORMAL	139	188	183	185	97	NORMAL
11	-	-	-	-	-	-	-	Sonuç Yok
12	T	NORMAL	139	188	183	185	97	NORMAL
13	-	-	-	-	-	-	-	Sonuç Yok
14	-	-	-	-	-	-	-	Sonuç Yok
15	T	NORMAL	148	155	183	185	97	NORMAL, ve Rekombinant
16	-	-	-	-	-	-	-	Sonuç Yok
17	T	NORMAL	139	188	183	185	97	NORMAL
18	T	NORMAL	139	188	183	189	99	NORMAL, ve Rekombinant
19	-	-	-	-	-	-	-	Sonuç Yok
20	-	-	-	-	-	-	-	Sonuç Yok
21	A	878-890 del ACATTAATGCTTC	148	155	187	189	99	MUTASYONLU
22	A	878-890 del ACATTAATGCTTC	148	155	187	189	99	MUTASYONLU
23	-	-	-	-	-	-	-	Sonuç Yok
24	A	878-890 del ACATTAATGCTTC	148	155	187	189	99	MUTASYONLU
25	A	878-890 del ACATTAATGCTTC	148	155	187	189	99	MUTASYONLU

PGT Vaka Performansı
STR ile Blastomer Analizi



Ardaşık Polar Body I - II ve Blastomer Analizi PGT Raporu

Embriyo #	Hücre Tipi	STR1	STR2	STR3	Gen	STR4	STR5	STR6	Öngörülen Oosit/Embriyo Genotipi	ET
1	PB1	120/158	115/103	215/207	N/Mutasyon	154/128	118/110	145/131	Oosit: NORMAL	Evet
	PB2	158	103	207	Mutasyon	128	110	131		
2	PB1	158	103	207	Mutasyon	128	110	131	Oosit: NORMAL* Embriyo: TAŞIYICI	Evet
	PB2	120	115	215	Normal	154	118	145		
	Blast	168/120	117/115	*207*/215	Mutasyon/N	146/154	ADO/118	*145*/*145*		
3	PB1	120/158	115/ADO	215/207	N/Mutasyon	154/ADO	118/110	145/131	Oosit: HASTA Embriyo: HASTA	Hayır
	PB2	120	115	215	Normal	154	118	145		
	Blast	168/158	117/*103*	*207*/*207*	Mutasyon/ Mutasyon	146/*128*	114/110	*145*/131		
4	PB1	120/158	115/103	215/207	N/Mutasyon	*128*/*128*	118/110	145/131	Oosit: YetersizVeri Embriyo: TRİZOMİ 15	Hayır
	PB2	AH	AH	AH	AH	AH	AH	AH		
	Blast	140/ 120/158	*103*/ 115/*103*	203/ 215/207	*N*/ *N*/Mutasyon	*128*/ 154/*128*	114/ 118/110	139/ *145*/131		
BABA		168/140	117/103	207/203	Mutasyon/ Normal	146/128	114/114	145/139	TAŞIYICI	
ANNE		120/158	115/103	215/207	Normal/ Mutasyon	154/128	118/110	145/131	TAŞIYICI	
ÇOCUK		168/158	117/103	207/207	Mutasyon / Mutasyon	146/128	114/110	145/131	HASTA	

N = Normal, AH = PCR Amplifikasyon Hatası, ADO = Allel Drop Out, *123* = Ebevenlerde aynı olan allel

* Homozigot PB I sonucu dolayısıyla Blastomer analizi ile devam edildi (PBI de muhtemel ADOI)

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Link STRs kullanarak Fragile-X Syndromu (FRAXA) Polar Body Analizi

Oosit #	Hücre Tipi	DXS297	DXS548	ATL1	Intron1	AC2	DXS8091	Oosit Genotip Tahmini	ET	İlave Bilgiler
1	PB1	Ado / 173	248 / 255	- / +	+ / -	179 / 178	Ado / 175	NORMAL	EVET	PB1 de ADO DXS297 ve DXS8091
	PB2	173	255	+	-	178	175			
2	PB1	173	255	+	-	178	175	NA	HAYIR	BB
	PB2	AH	AH	AH	AH	AH	AH			
3	PB1	167 / 173	248 / 255	- / +	+ / -	179 / 178	181 / 175	NORMAL	EVET	
	PB2	173	255	+	-	178	175			
4	PB1	167 / 173	255	+	-	178	175	NORMAL *	EVET	REKOMBİNANT DXS297 sonrası
	PB2	173	248	-	+	179	181			
5	PB1	167 / 173	248 / 255	- / +	+ / -	179 / 178	181 / 175	NORMAL	EVET	ATL1 için PB2 de AH
	PB2	173	255	FA	-	178	175			
6	PB1	167 / 173	248 / 255	Ado/ +	+ / -	179 / Ado	181 / 175	NORMAL	EVET	PB1 de ADO ATL1 ve AC2
	PB2	173	255	+	-	178	175			
7	PB1	167	248	-	+	179	181	HASTA *	HAYIR	
	PB2	173	255	+	-	178	175			
8	PB1	167 / 173	248 / 255	- / +	+ / -	179 / 178	181 / 175	HASTA	HAYIR	
	PB2	167	248	-	+	179	168			
		167 / 173	248 / 255	- / +	+ / -	179 / 178	181 / 175	ANNE (TAŞIYICI)		
		167	249	+	-	178	173	BABA		
		173	255	+	-	178	175	ÇOCUK (HASTA)		

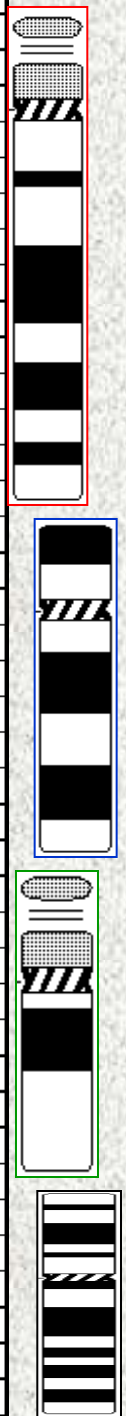
PB1 – POLAR BODY 1; PB2 – POLAR BODY 2

ADO - Allele Drop Out; AH – Amplificasyon Hatası; **ET** – Embryo Transfer; **NA** – Not Applicable; **BB** – Blastomer Biopsisi

Kırmızı renkli alleler mutant kromozom üzerindedir; siyah renkli alleller normal kromozom üzerindedir**.**

* PB1 de olası ADO sebebiyle % 2 den daha az yanlış tanı şansı.

STR	FATHER	MOTHER
D13S1267	75/75	75/75
D13S742	**257/257**	**260/278**
D13S1303	**132/148**	**137/150**
D13S813	183/186	186/192
D13S1236	95/99	91/96
D13S1493	242/250	242/250
D13S1317	139/151	137/137
D13S175	125/125	125/125
D13S317	211/215	211/211
D13S174	**123/129**	**119/123**
D13S1282	131/140	130/130
D13S284	191/197	192/196
D13S1275	132/139	136/139
D13S800	286/290	286/302
D18S1145	129/131	129/129
D18S463	120/130	118/130
D18S56	**135/137**	**133/135**
D18S1144	**114/116**	**98/102**
D18S1127	145/148	136/150
D18S57	108/108	100/100
D18S66	**151/153**	**148/157**
D18S386	360/363	349/349
D18S1124	127/154	126/146
D18S61	134/152	155/155
D21S1903	154/156	154/154
D21S3	276/279	279/283
D21S1888	**141/165**	**145/154**
D21S1411	190/190	182/186
D21S1899	136/143	132/138
D21S70	**187/197**	**187/201**
D21S11	221/226	221/232
D21S910	**164/197**	**191/205**
D21S268	134/137	126/134
DXS1215	**175**	**175/179**
DXS1055	110	105/105
DXS8090	166	161/166
DXS1068	127	131/131
DXS1684	**151**	**139/151**
DXS8025	**190**	**186/190**



Kromozom
Anöploidi
Testi

STR ile

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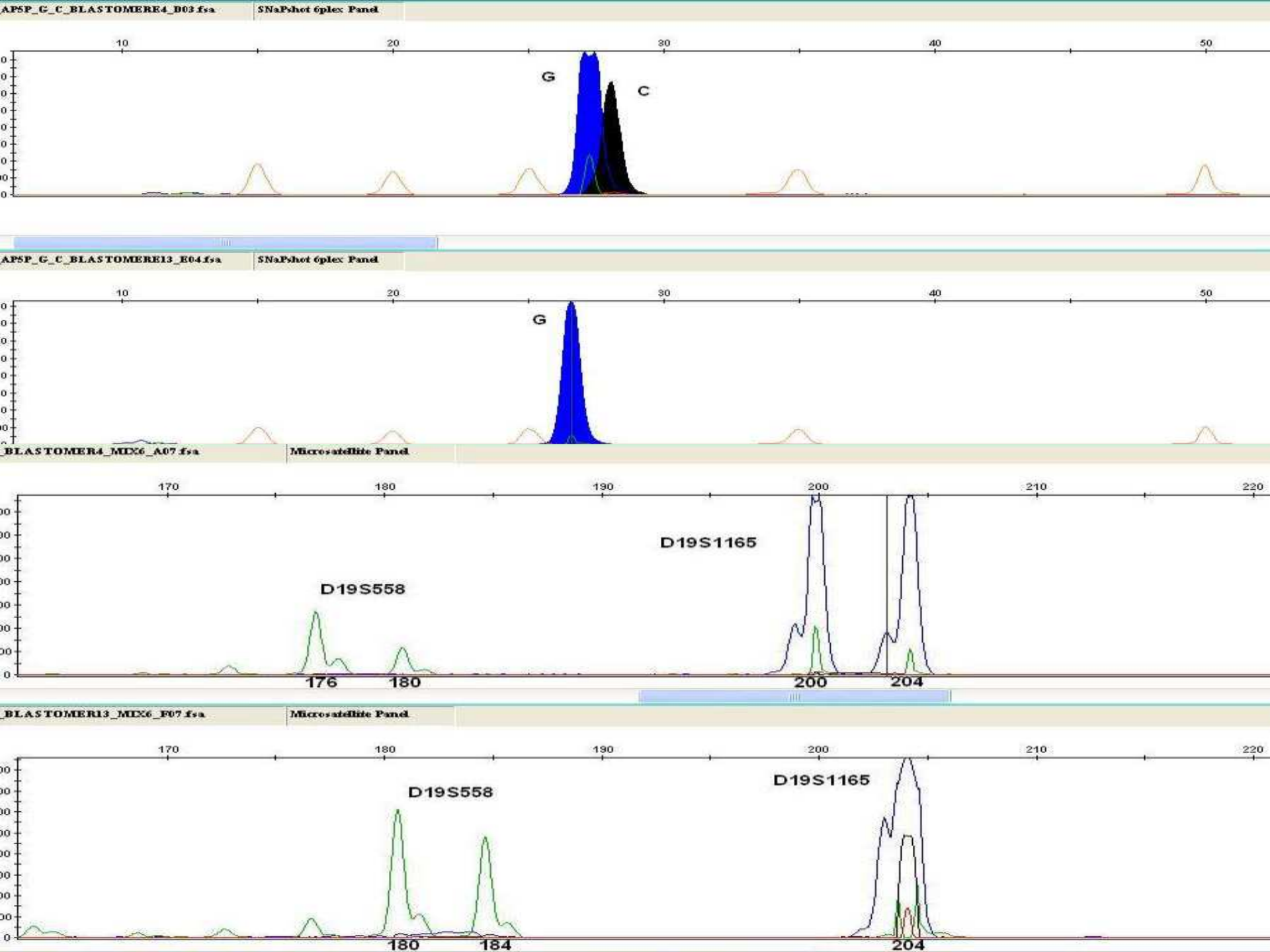
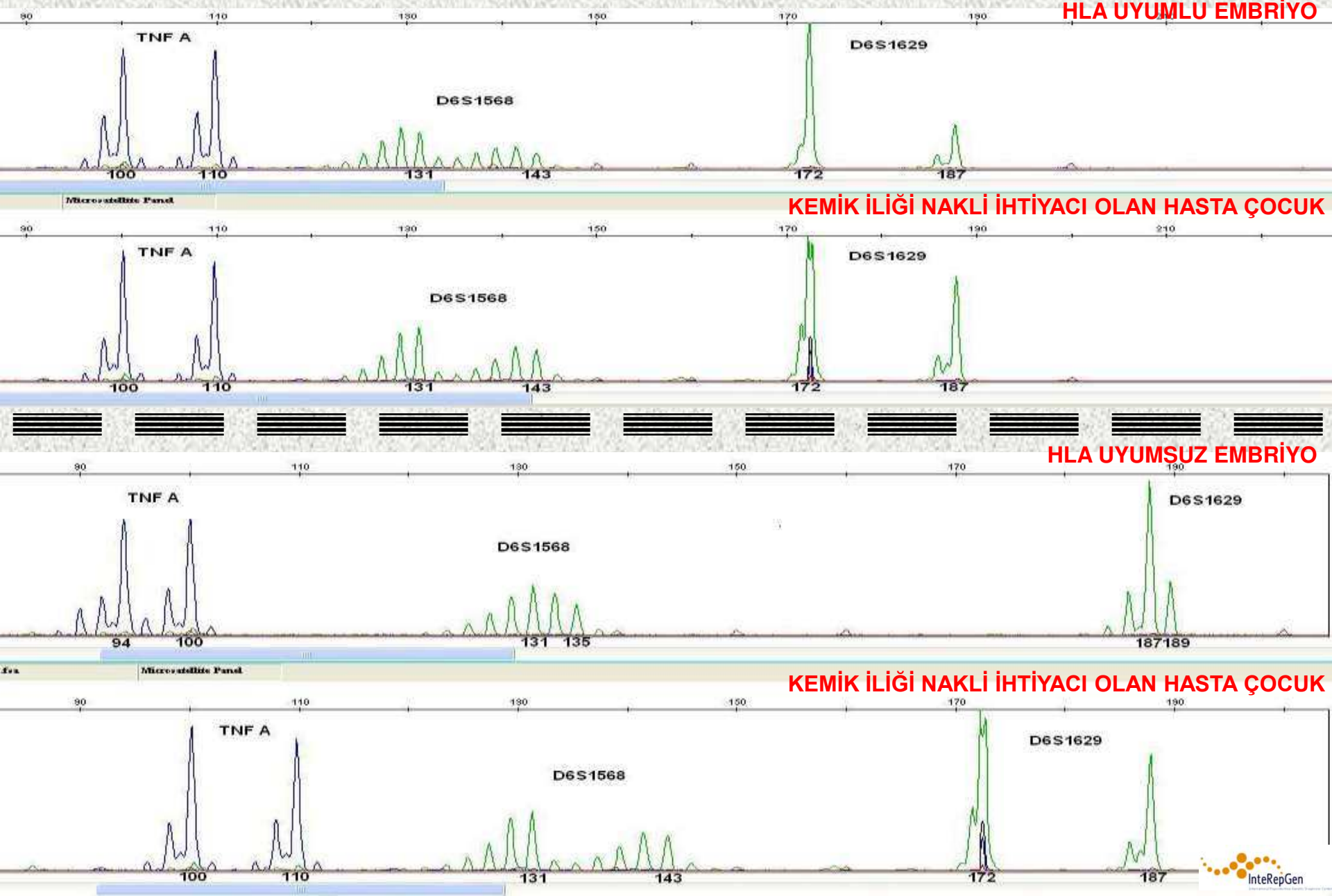

AP5P_G_C_BLASTOMERE4_D03.fsa
SNaPshot 6plex Panel
10
20
30
40
50
G
C
AP5P_G_C_BLASTOMERE13_E04.fsa
SNaPshot 6plex Panel
10
20
30
40
50
G
_BLASTOMER4_MIX6_A07.fsa
Microsatellite Panel
170
180
190
200
210
220
D19S1165
D19S558
176
180
200
204
_BLASTOMER13_MIX6_F07.fsa
Microsatellite Panel
170
180
190
200
210
220
D19S558
D19S1165
180
184
204

PGT HLA Genotiplemesi



PGT YAPILAN HASTALIKLAR

A. NÖROMUSKULER HASTALIKLAR

Fragile X mental retardation syndrome (FMR1/ FRAXA locus), Xq27.3 (RNA-binding protein FMRP, *FMR1*; 309550).
Duchenne muscular dystrophy (DMD), Xp21.2 (dystrophin, *DMD*; 300377).
Myotonic dystrophy-1 (DM1), 19q13 (dystrophia myotonica protein kinase, *DMPK*; 605377)
Emery-Dreifuss muscular dystrophy (EDMD2), 1q21.2 (lamin A/C, *LMNA*; 150330).
Facioscapulohumeral muscular dystrophy (FSHD), 4q35 (*FRG1*, (601278),
Myotubular myopathy-1 X linked (MTM1), Xq28 (myotubularin, *MTMR1*; 300171).
Spinal muscular atrophy type I (SMA I), 5q12-q13 (*SMN1*; 600354).
Huntington disease (HD), 4p16.3 (huntingtin, *IT15*, +143100)
Hereditary Amyloid Polyneuropathy, 18q11.2-q12.1 (thyroxine binding prealbumin / transthyretin, TTR1, +176300)
Familial dysautonomia (FD) (or HSAN3), 9q31 (IKK complex associated protein – IKAP, *IKBKAP* (603722).
Charcot-Marie-Tooth disease type 1A (CMT1A), 17p11.2 (peripheral myelin protein-22, *PMP22*; 601097).
Charcot-Marie-Tooth disease type 1B (CMT1B), 1q22 (myelin protein zero, *MPZ*; 159440).
Charcot-Marie-Tooth disease X-linked (CMTX1), Xq13.1 (connexin 32 , *GJB1*: 304040).
Charcot-Marie-Tooth disease type 2E (CMT2E), 8p21 (light polypeptide neurofilament protein/NEFL, *NFL*; 162280).
Friedreich ataxia (FRDA1), 9q13 (frataxin, *FXN*; 606829),
X-linked hydrocephalus (HYCX or HSAS), Xq28 (L1 cell adhesion molecule, *L1CAM*; 308840).
Leigh syndrome (infantile subacute necrotizing encephalopathy - SNE), 9p34 (*SURF1*, 185620).
Norrie disease (ND), Xp11.4 (norrin, *NDP*; 300658).
Pelizaeus-Merzbacher disease (PMD), Xq22 (proteolipid protein-1, *PLP1*; 300401).
Rett syndrome (RTT), Xq28 (methyl-CpG-binding protein-2, *MECP2*; 300005).
Spinocerebellar ataxia-1 (SCA1), 6p23 (ataxin-1, *ATXN1*; 601556).
Spinocerebellar ataxia-2 (SCA2), 12q24 (ataxin-2, *ATXN2*; 601517).
Spinocerebellar ataxia-3 (SCA3 - Machado-Joseph disease), 14q24-q31 (ataxin-3 gene, *ATXN3*; 607047).
Spinocerebellar ataxia-6 (SCA6), 19p13 (*CACNA1A*; 601011).
Spinocerebellar ataxia-7 (SCA7), 3p21-p12 (ataxin-7, *ATXN7*; 607640).
Holoprosencephaly-3 (HPE-3), 7q36 (human sonic hedgehog homolog, *SHH*; 600725).
Torsion dystonia (DYT1), 9q34.1 (torsin-A, *DYT1*; 605204).
Tuberous sclerosis complex -1 (TSC1), 9q34 (hamartin, *TSC1*; 605284)
Tuberous sclerosis complex -2 (TSC2), 16p13 (tuberin, *TSC2;* 191092),

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B. METABOLİK HASTALIKLAR

Tay-Sachs disease (TSD), 15q23-q24 (hexosaminidase A gene, *HEXA*; 606869).
Krabbe Disease (Globoid Cell Leukodystrophy – GLD), 14q31 (galactosylceramidase gene, *GALC*; 606890).
Gaucher disease Type 1 (GD I), 1q21 (acid beta-glucosidase, *GBA*; 606463).
Metachromatic leukodystrophy (MLD), 22q13.31 (arylsulfatase A gene, *ARSA*; 607574).
Adrenoleukodystrophy (ALD), Xq28 (an ATPase binding cassette protein/peroxisomal transporter, *ABCD1*; 300371).
Neuronal ceroid lipofuscinosis-2 (CLN2), 11p15.5 (a lysosomal peptidase, *CLN2*; 607998).
Citrullinemia (CTLN1), 9q34.1 (argininosuccinate synthetase, *ASS*; 603470).
Fabry disease, Xq22 (alpha-galactosidase A, GLA; 300644).
GM1-gangliosidosis, 3p21.33 (beta-galactosidase-1, *GLB1*; 611458).
GM2 - gangliosidosis type II (Sandhoff disease), 5q13 (beta subunit of hexosaminidase, *HEXB*; 606873).
Mucopolysaccharidosis II (MPS2, Hunter Syndrome), Xq28 (iduronate sulfatase, *IDS*; +309900)
Mucopolysaccharidosis type IH (Hurler syndrome), 4p16.3 (alpha-L-iduronidase, *IDUA*; 252800).
Hypophosphatasia, 1p36.1-p34 (tissue-nonspecific alkaline phosphatase, *ALPL*; 171760).
X-linked hypophosphatemia (XLH; HYP), Xp22.2-22.1 (phosphate-regulating endopeptidase, *PHEX*; 300550)
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD), 2p23 (*HADHA*; 600890).
Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD), 1p31 (*ACADM*; 607008).
Methylenetetrahydrofolate reductase deficiency (MTHFR), 1p36.3 (*MTHFR*; 607093).
Ornithine carbamoyltransferase deficiency (OTC), Xp21.1 (*OTC*; 300461).
Propionic academia, 13q32 (propionyl-CoA carboxylase, *PCCA*; 232000)
Succinic semialdehyde dehydrogenase (SSADH), 6p22 (*ALDH5A1*; 610045).
Alpha-mannosidosis (LAMAN), 19p13.2-q12 (MAN2B1; 609458).
Crigler-Najjar syndrome (CNS), 2q37 (UGT1A1; 191740).

C. BÖBREK HASTALIKLARI

Polycystic kidney disease 1 - autosomal dominant (PKD1), 16p13 (*PKD1*; 601313)
Polycystic kidney disease 2 - autosomal dominant (PKD 2), 4q21-q23 (*PKD2*; 173910)
Polycystic kidney disease - autosomal recessive (ARPKD), 6p21-p12 (fibrocystin, *PKHD1*; 606702).
Alport syndrome (ATS), Xq22.3 (alpha-5 chain of basement membrane collagen, *COL4A5*; 303630).
Congenital nephrotic syndrome (and Pierson syndrome), 3p21 (laminin beta-2, *LAMB2*; 150325)
Nephropathic cystinosis (CTNS), 17p13 (cystinosin, *CTNS*; 606272).
Zellweger syndrome, 8q21 (peroxisomal membrane protein 3 –PXMP3, peroxin-2, *PEX2*;

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D. HEMATOLOJİK HASTALIKLAR

Thalassemia-alpha, 16pter-p13.3 (alpha globin, *HBA1;* +141800)
Thalassemia-beta, 11p15.5 (beta-globin, HBB; +141900)
Sickle cell anemia, 11p15.5 (beta-globin, HBB; +141900)
Fanconi anemia A (FAA), 16q24.3 (complementation group A, *FANCA;* 607139).
Fanconi anemia C (FAC), 9q22.3 (complementation group C, *FANCC*; 227645).
Fanconi anemia F (FA-F), 11p15 (complementation group F, *FANCF*; 603467).
Fanconi anemia J (FA-J), 17q22 (complementation group J, *FANCJ*; 605882),
Diamond-Blackfan anemia (DBA), 19q13.2 (ribosomal protein S19, *RPS19*; 603474).
Hemophilia A (HEMA), Xq28 (coagulation factor VIII, F8; +306700)
Hemophilia B (HEMB), Xq27 (plasma thromboplastin component (PTC)/ coagulation factor VIII, F9; +306900)
Hoyeraal-Hreidarsson syndrome (HHS), Xq28 (dyskerin, *DKC1*; 300126)
Kell blood group (KEL), 7q33 (Kell glycoprotein, *KEL*; +110900)

E. İMMÜN YETMEZLİK HASTALIKLARI

Immunodeficiency with hyper-IgM (HIGM1, X-linked hyper-IgM syndrome), Xq26 (CD40 ligand, *CD40LG*; 300386).
Omenn syndrome (severe combined immunodeficiency with hyper eosinophilia), 11p13 (*RAG1*; 179615)
Wiskott-Aldrich syndrome (WAS), Xp11 (*WAS*; 300392).
Ataxia-telangiectasia (AT),11q23.3(phosphatidylinositol-3 kinase, ataxia-telangiectasia mutated gene, *ATM*; 607585).

F. TÜMÖRAL HASTALIKLAR

Familial adenomatous polyposis (FAP/ or APC), 5q21-q22 (*APC*; 175100),
Hereditary nonpolyposis colorectal cancer type 1 (HNPCC1), 2p22-p21 (*MSH2*; 609309),
Hereditary nonpolyposis colorectal cancer type 2 (HNPCC2), 3p21.3 (*MLH1*; (120436),
Retinoblastoma (RB), 13q14 (*RB1*; +180200)
Nevoid basal cell carcinoma syndrome (BCNS, Gorlin Syndrome), 9q22.3 (*PTCH1*; 601309).
Li-Fraumeni syndrome-1 (LFS-1), 17p13.1 (p53, *TP53*; 191170).
Neurofibromatosis type 1 (NF1), 17q11.2 (neurofibromin, *NF1*; +162200)
Neurofibromatosis type II (NF2), 22q12.2 (neurofibromin-2 /or merlin, *NF2*; 607379).
von Hippel-Lindau syndrome (VHL), 3p26-p25 (*VHL*; 608537).

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G. DİĞER HASTALIKLAR

Cystic fibrosis, 7q31 (Cystic fibrosis transmembrane conductance regulator, *CFTR*; 602421)
Crouzon syndrome (Craniofacial Dysostosis type 1/CFD1), 10q26 (fibroblast growth factor receptor-2, *FGFR2*, 176943).
Deafness, nonsyndromic neurosensory – autosomal recessive (DFNB1), 13q11-q12 (connexin-26, *GJB2*; 121011)
Popliteal pterygium syndrome (PPS), 1q32-q41 (interferon regulatory factor-6, *IRF6*; 607199).
Smith-Lemli-Opitz syndrome (SLOS), 11q12-q13 (sterol delta-7-reductase, *DHCR7*; 602858),
Currarino syndrome, 7q36 (homeobox gene, *HLXB9*; 142994).
Hereditary pancreatitis (PCTT), 7q35 (cationic trypsinogen, *PRSS1*; 276000)
Darier-White disease (DAR), 12q23-q24.1 (SERCA2 Ca(2+)-ATPase, *ATP2A2*; 108740).
Hypohidrotic ectodermal dysplasia X-linked (ED1), Xq12-q13.1 (ectodysplasin-A, *EDA*; 300451).
Hypohidrotic ectodermal dysplasia (HED), 2q11-q13 (ectodysplasin anhidrotic receptor gene, *EDAR*; 604095)
Epidermolysis bullosa dystrophica autosomal recessive (EBR1), 3p21.3 (type VII collagen, *COL7A1*; 120120).
Epidermolysis bullosa letalis – junctional, 1q32 (beta-3 part of laminin-5, *LAMB3*; 150310).
Epidermolysis bullosa simplex, 8q24 (Plectin 1, *PLEC1*, 601282).
Incontinentia pigmenti (IP), Xq28 (IKK-gamma, *IKBKG* (or *NEMO*); 300248).
Marfan syndrome (MFS), 15q21.1 (fibrillin-1, *FBN1*; 134797),
Oculocutaneous albinism type IA (OCA1A), 11q14-q21 (tyrosinase, *TYR*; 606933).
Oculocutaneous albinism type II (OCA2), 15q11.2-q12 (integral melanosomal membrane protein, *OCA2* (611409).
Ocular albinism type 1(OA1), Xp22.3 (*GPR143 (or OA1)*; +300500)
Optic atrophy-1 (OPA1), 3q28-q29 (*OPA1*; 605290)
Choroideremia (CHM), Xp21.2 (Rab escort protein-1, *REP1*; 300390).
Blepharophimosis, ptosis, and epicanthus inversus syndrome(BPES), 3q23 (forkhead transcription factor, *FOXL2*; 605597).
Stickler syndrome (STL1), 12q13 (*COL2A1* ; 120140).
Osteogenesis imperfecta (OI), 17q21-q22 (*COL1A1*; 120150)
Osteopetrosis Infantile malignant autosomal recessive (OPTB1), 11q13, (*TCIRG1*; 604592)
Multiple exostoses type I (EXT1), 8q24 (exostosin-1, *EXT1*; 608177),
Multiple synostoses syndrome (SYNS1 -Facioaudiosymphalangism), 17q22 (noggin, *NOG*; 602991)
Treacher Collins syndrome (TCS), 5q32-q33 (*TCOF1*; 606847).
Brachydactyly type B (BDB1), 9q22 (receptor tyrosine kinase-like orphan receptor 2, *ROR2*; 602337).
Robinow syndrome-Autosomal recessive, 9q22 (receptor tyrosine kinase-like orphan receptor 2, *ROR2*; 602337).
Cockayne Syndrome Type B, 10q11 (group 6 excision-repair cross-complementing protein, ERCC6: 609413).

H. HLA HAPLOTİPLEMESİ

PGT

- Prenatal Tanıya alternatif değildir
  Avantajları ve Üstünlükleri
- Tekrarlanabilir protokoller
  Güvenilirlik Oranı ve Başarı Devamlılığı
- Takım Çalışması
  Genetik Danışma ve Genetik Lab.

SÜREKLİLİK