Диагностика геномных и хромосомных аномалий у детей методом сравнительной геномной гибридизации

Генетические школы
genschool.ru

Лекция прочитана в рамках проекта genschool.ru,
октябрь 2016 г.

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ФГБУ НЦАГиП им. В.И.Кулакова Минздрава России
Москва, 2016.

План доклада

- 1. Традиционные методы цитогенетики. Достоинства и недостатки

  2. Общие принципы метода aCGH.

  3. Особенности работы разных платформ:

     a. Agilent

     b. Affymetrix

     c. Illumina

  4. Общее обзорное сравнение платформ.

  5. Разбор клинических случаев.

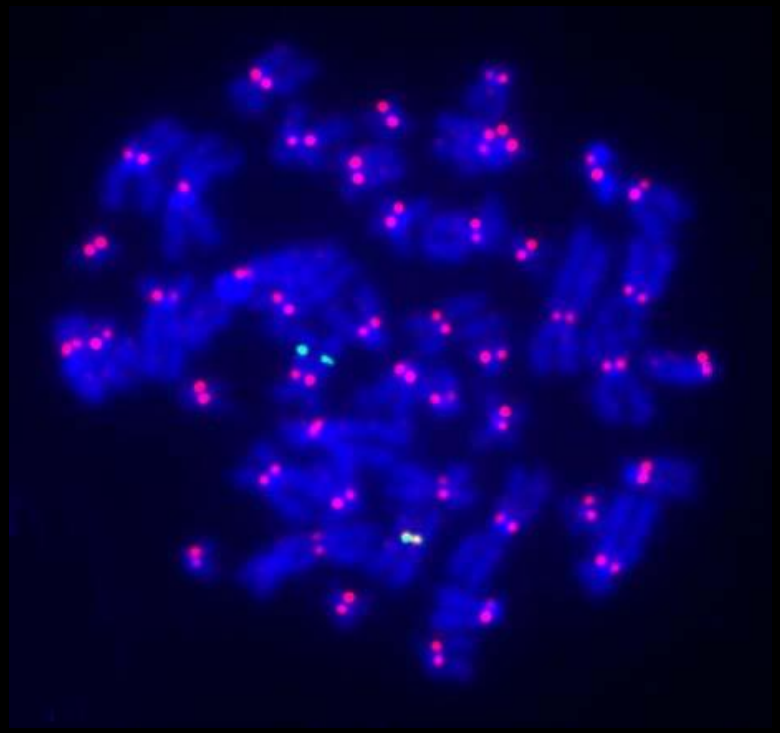
  6. Рекомендации по выбору метода исследования в зависимости от поставленной задачи.

  7. Важность оформления сопроводительной документации для проведения анализа. Взаимодействие клиники и лаборатории.

Традиционные методы цитогенетики.

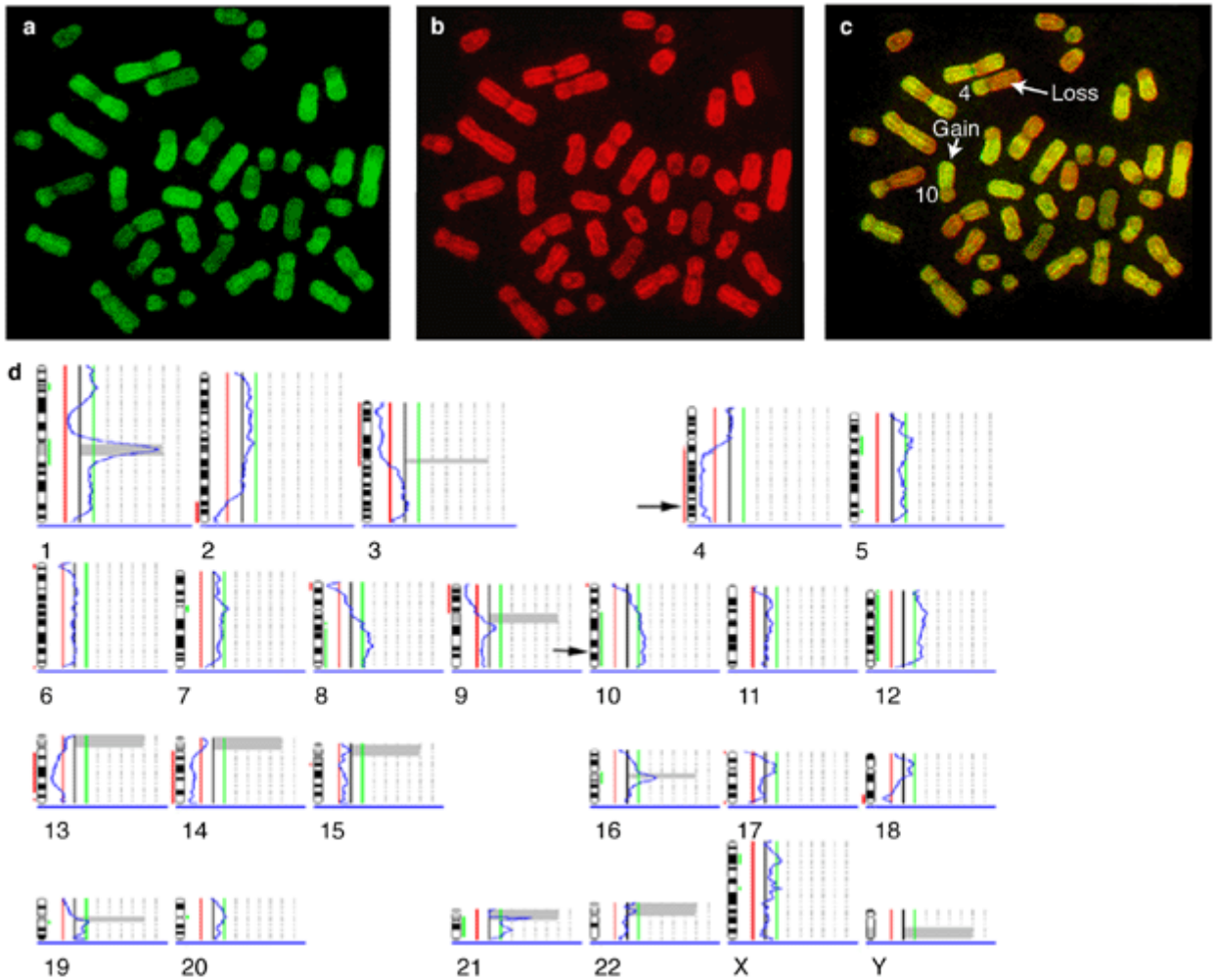
Кариотипирование

FISH



- Сравнительная геномная гибридизация на биологических чипах
- array Comparative Genome Hybridisation (aCGH)
- Молекулярное кариотипирование
- Виртуальное кариотипирование

Общие принципы метода aCGH.



Comparative genomic hybridisation (CGH) analysis of a lymph node metastasis from a renal cell carcinoma

Expert Reviews in Molecular Medicine © 2000 Cambridge University Press

Области применения aCGH

- Полногеномное профилирование
- Выявление CNV
- Генотипирование (SNP arrays)
- Исследование экспрессии генов
- Изучение митохондриальной ДНК
- Изучение микроРНК
- Изучение метилирования

Agilent

Agilent Technologies

Agilent Technologies
SureScan Microarray Scanner

Agilent Two-Color Competitive Hybridization aCGH Method

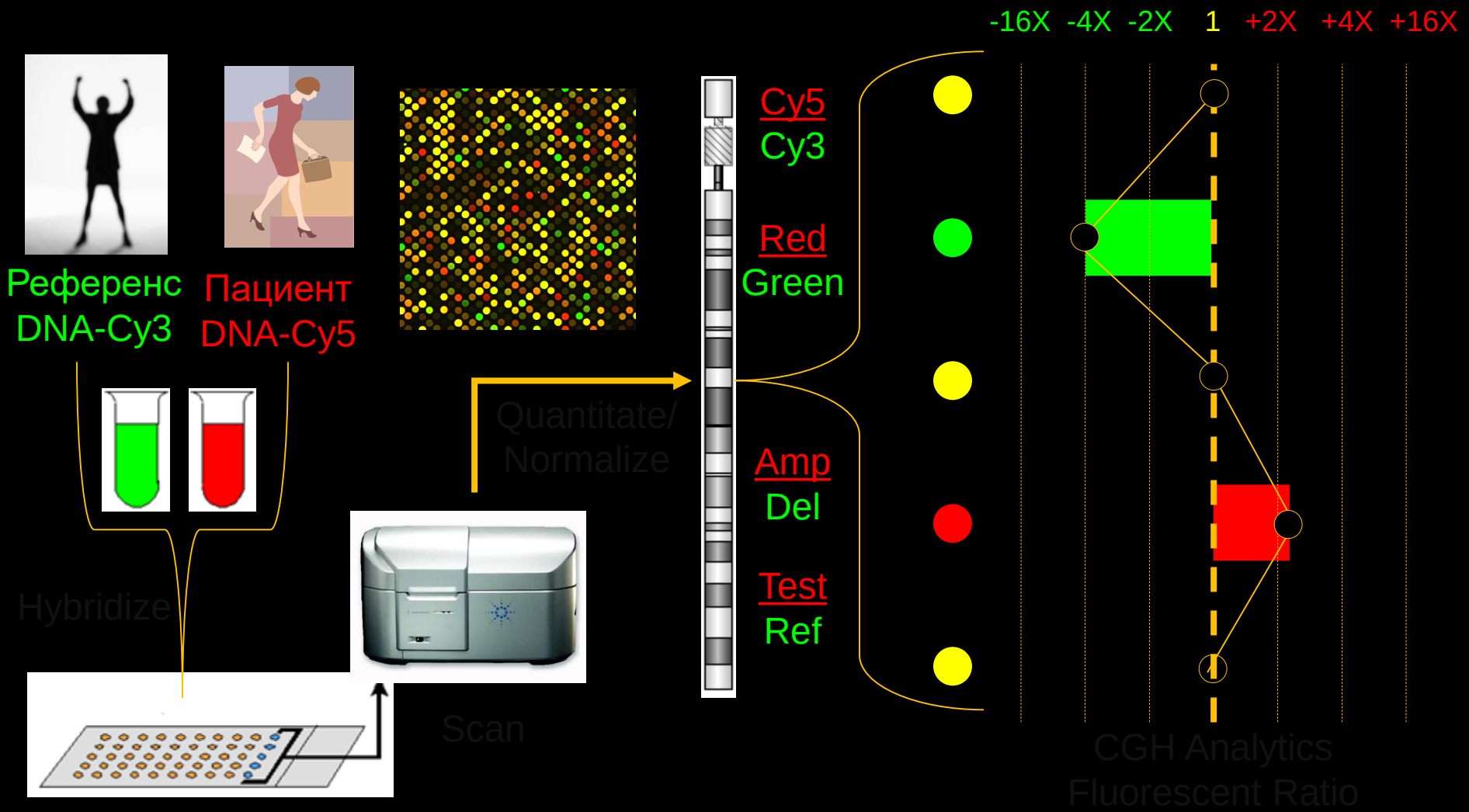


Схема aCGH

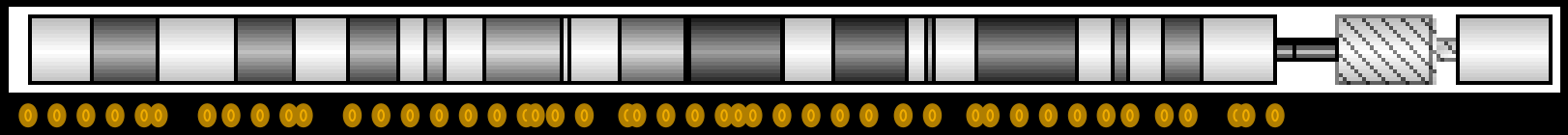


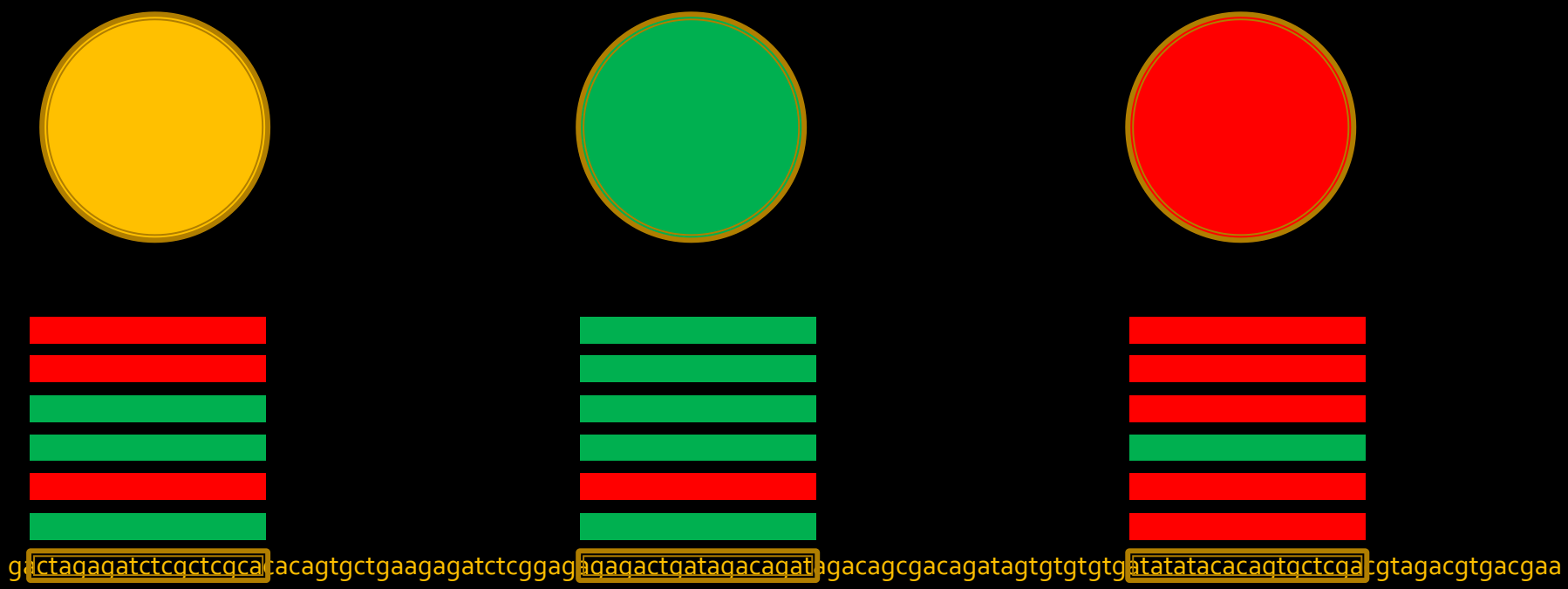
Схема aCGH

gactagagatctcgctcgcacacagtgctgaagagatctcggagagagactgatagacagatagacagcgacagatagtgtgtgtatatatacacagtgctcgacgtagacgtgacgaa

Схема aCGH

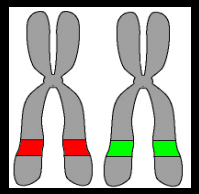
делеция

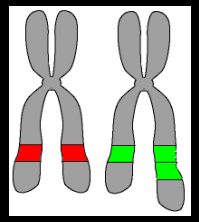
дупликация

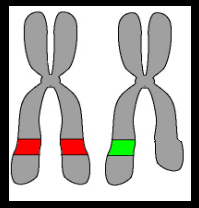


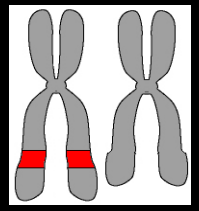
Измерения CNV

Копийность референсного аллеля	Копийность исследуемого аллеля	Отношение	Логарифм
2	2	1	0,0
2	3	1,5	0,6
2	1	0,5	-1,0
2	0	0	-∞

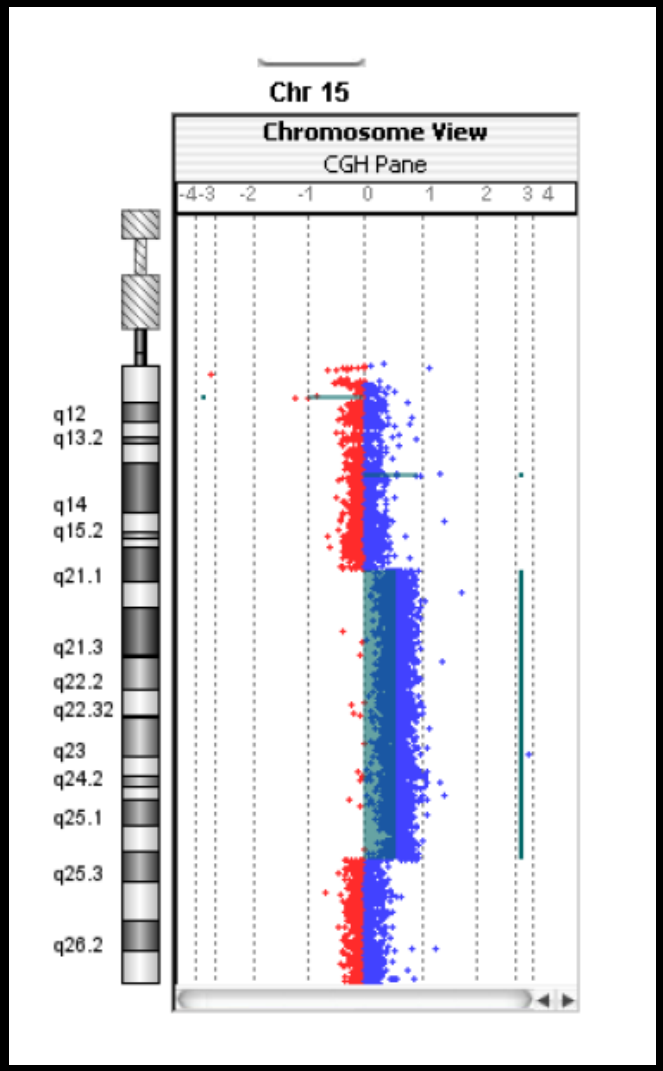


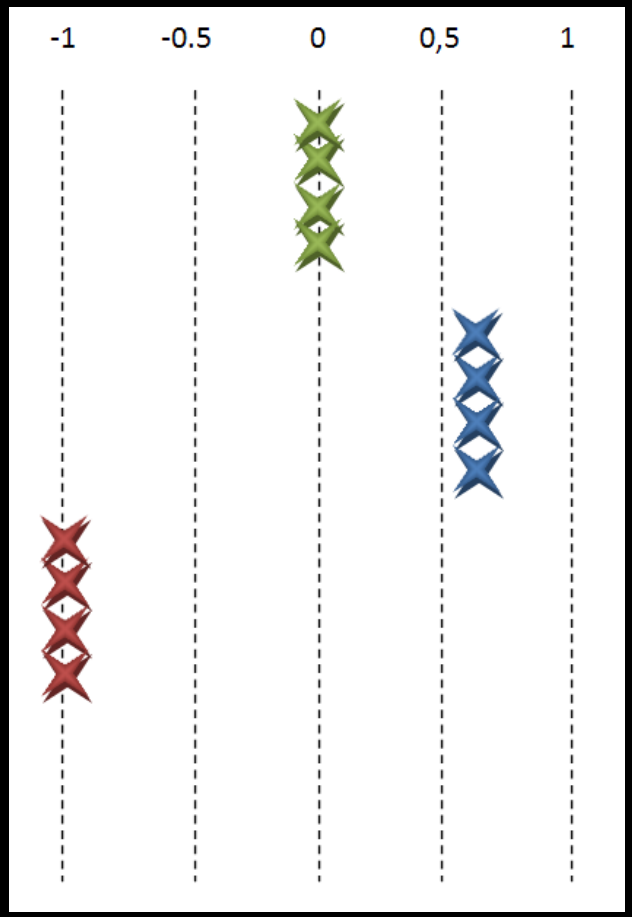


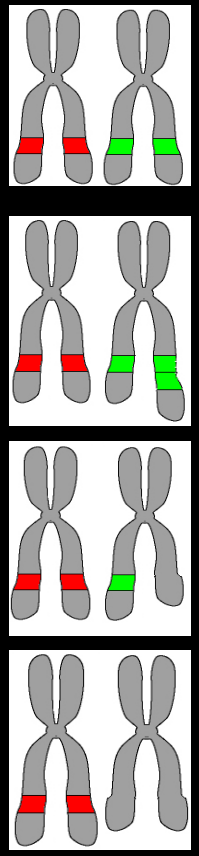




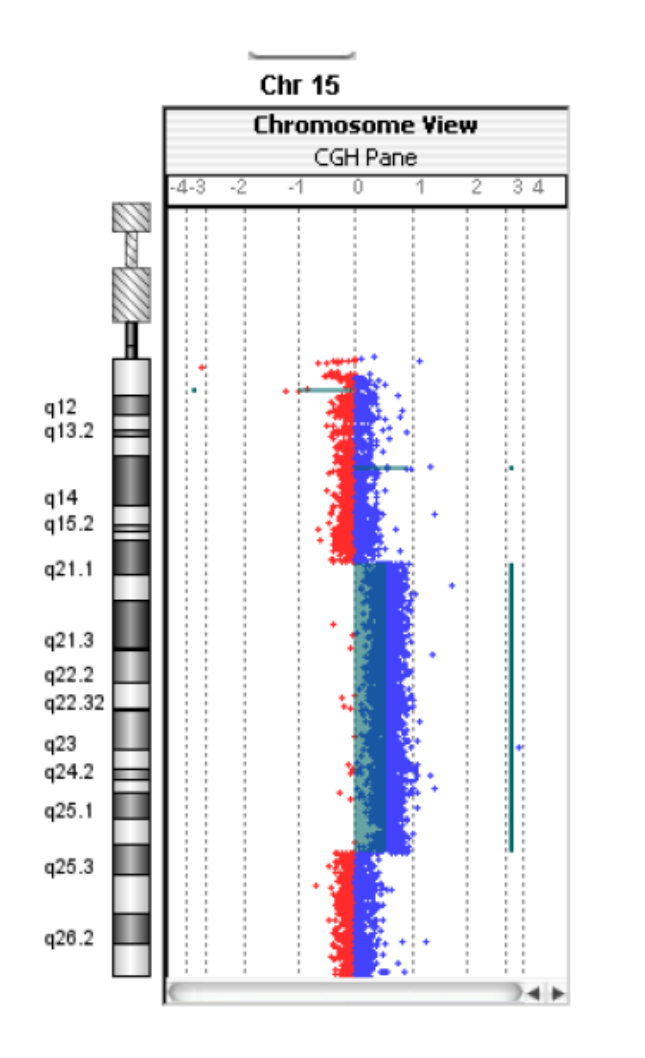
Измерения CNV

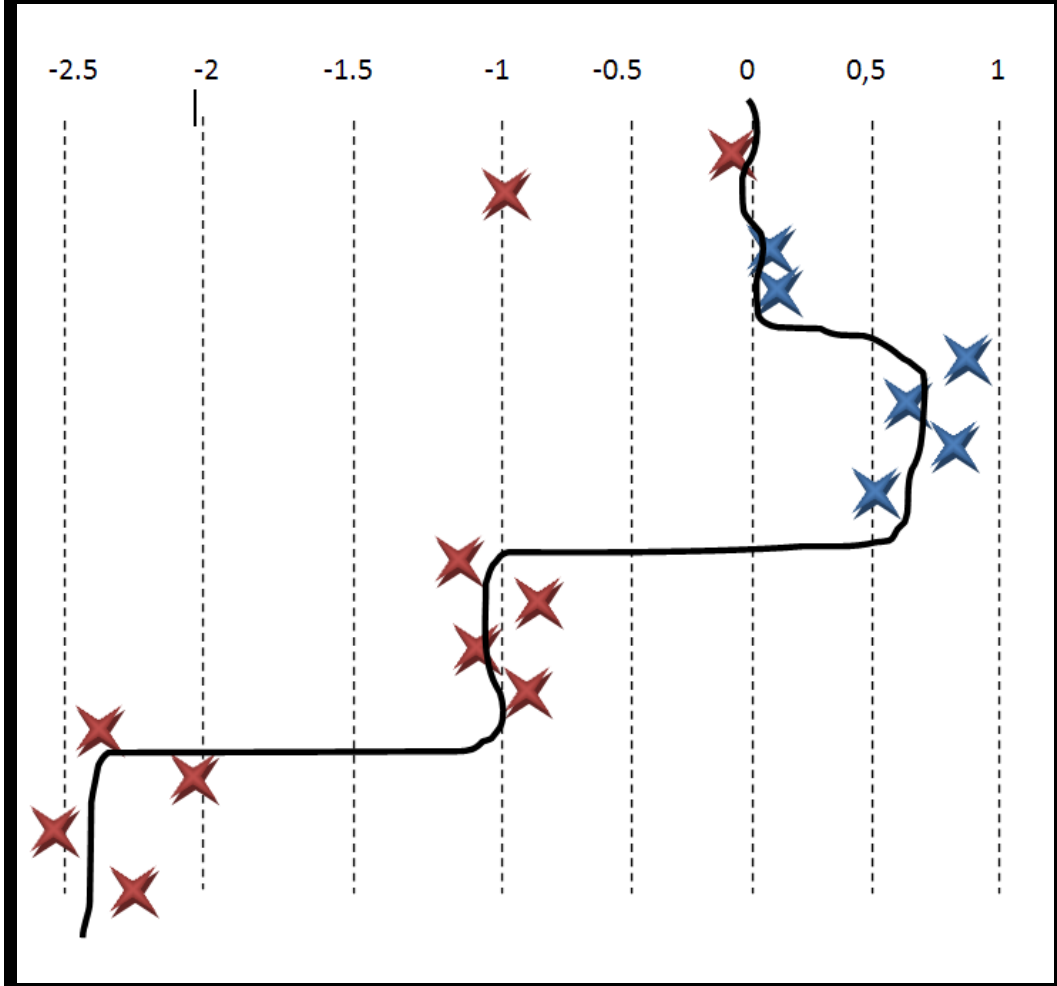


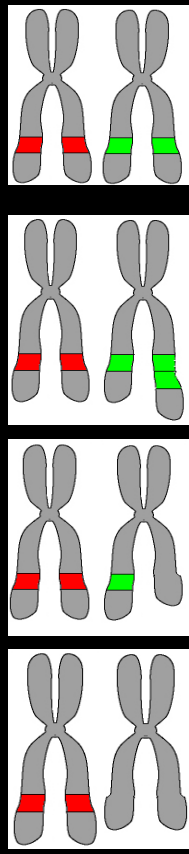




Измерения CNV



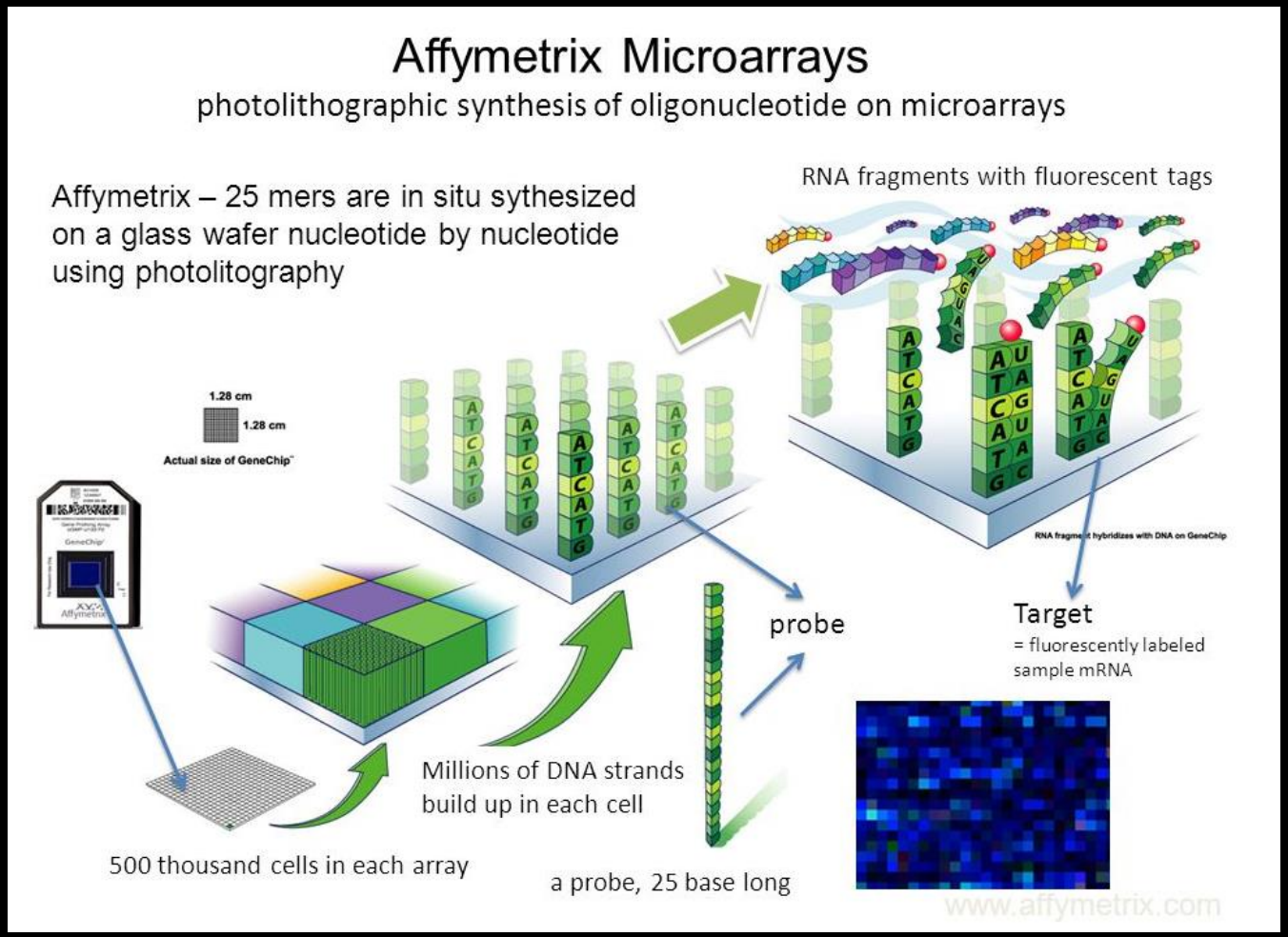




Affimetrix

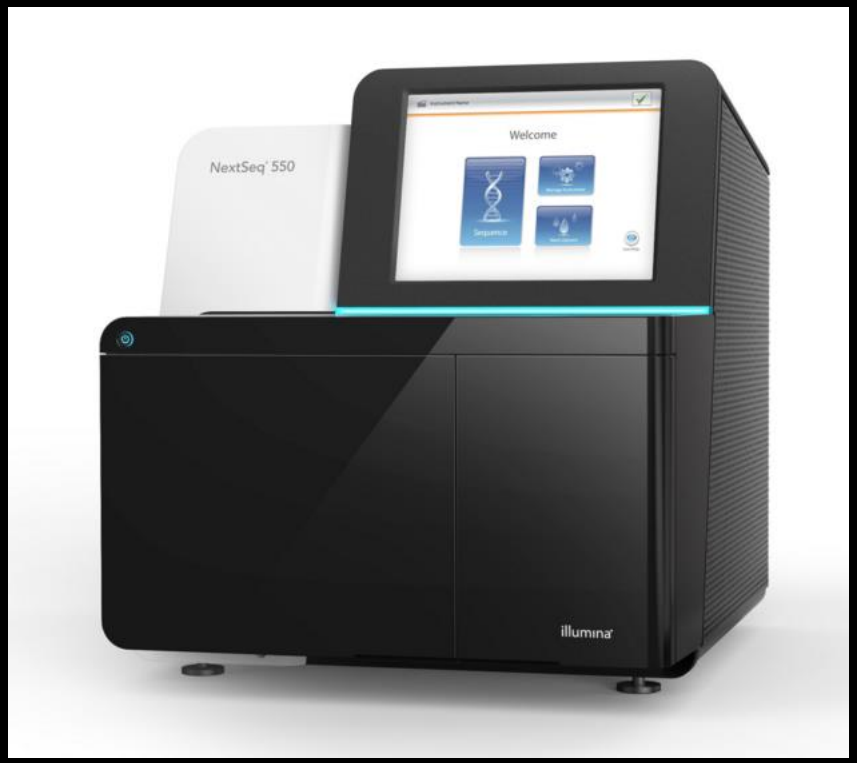
Affymetrix GeneChip® Scanner 3000 7G

Affimetrix


Affymetrix Microarrays
photolithographic synthesis of oligonucleotide on microarrays
Affymetrix – 25 mers are in situ sythesized on a glass wafer nucleotide by nucleotide using photolitography
RNA fragments with fluorescent tags
1.28 cm
1.28 cm
Actual size of GeneChip
probe
Target
= fluorescently labeled sample mRNA
Millions of DNA strands build up in each cell
500 thousand cells in each array
a probe, 25 base long
www.affymetrix.com

Illumina

Illumina iScan

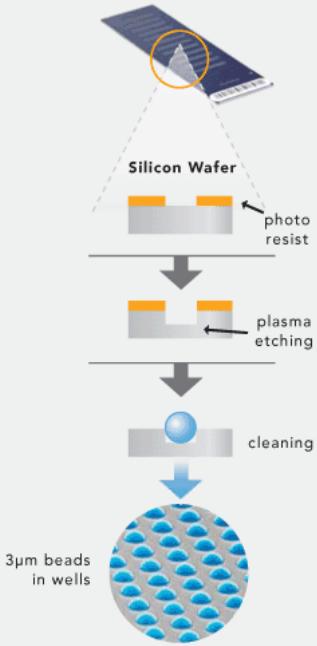


NextSeq 550 System

Illumina

Figure 1: HumanOmni5Exome-4 BeadChip

With over 4.5 million markers, including tag SNP coverage down to 1% MAF and functional exonic content, the four-sample HumanOmni5Exome-4 BeadChip is the ideal array for whole-genome genotyping and CNV analysis.



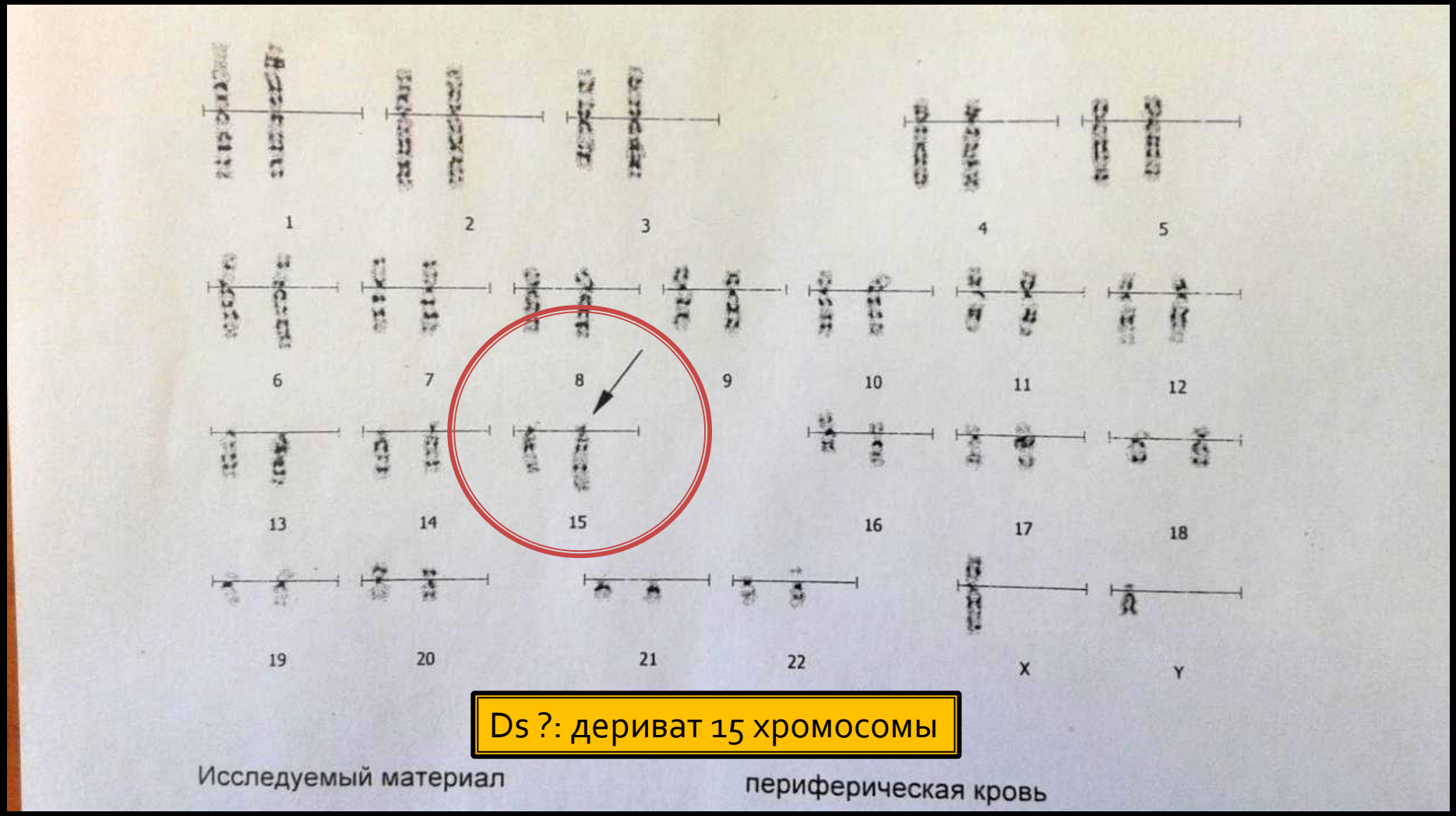
Illumina multi-sample array formats

Общее обзорное сравнение платформ.

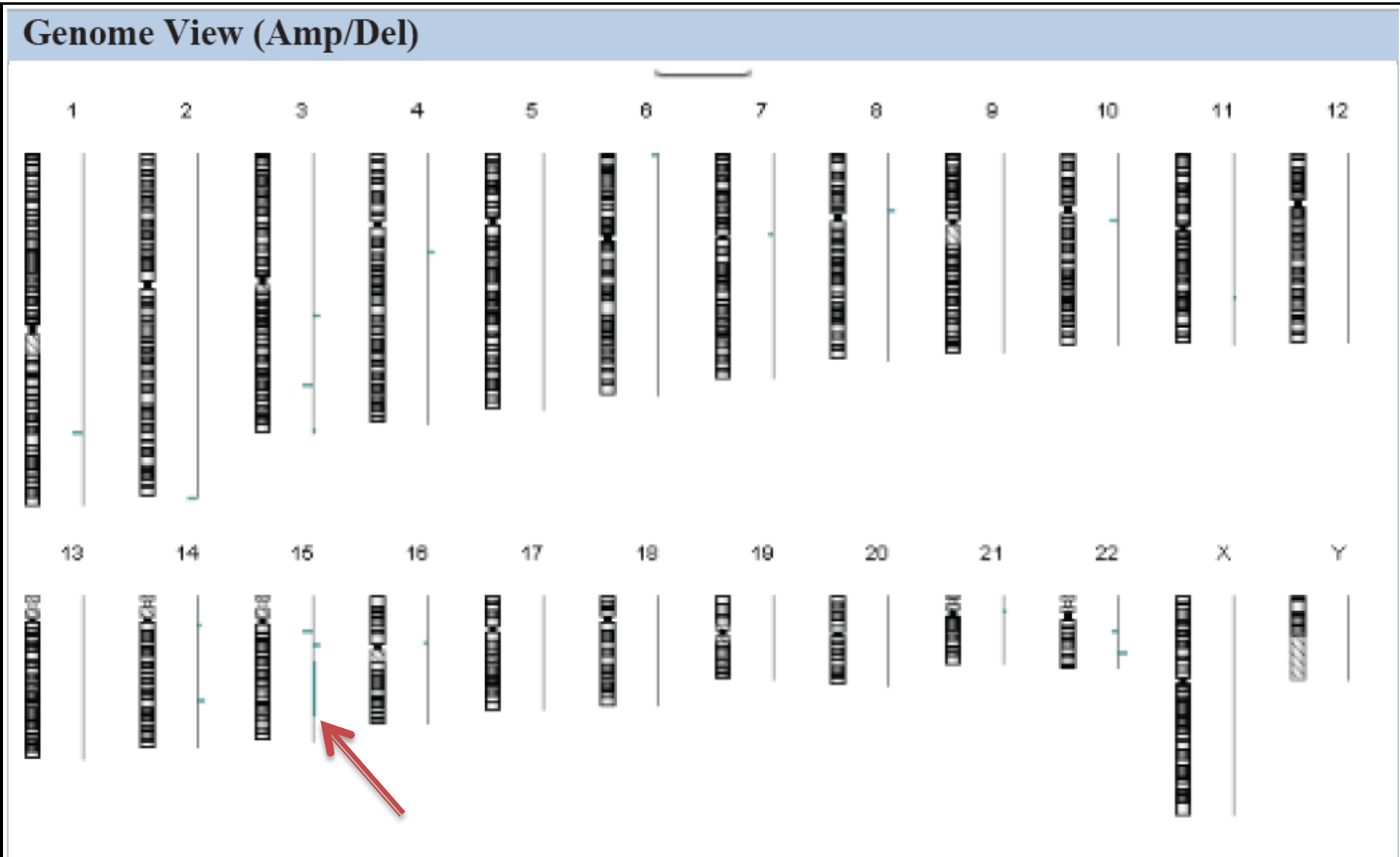
- Наличие зондов в интересующей области
- Плотность зондов
- Наличие и количество реплик зондов
- Возможность выявления SNP и/или CNV.
- Возможность выявления однородительской дисомии/потери гетерозиготности
- Стоимость анализа
- Время выполнения анализа

Разбор клинических случаев.

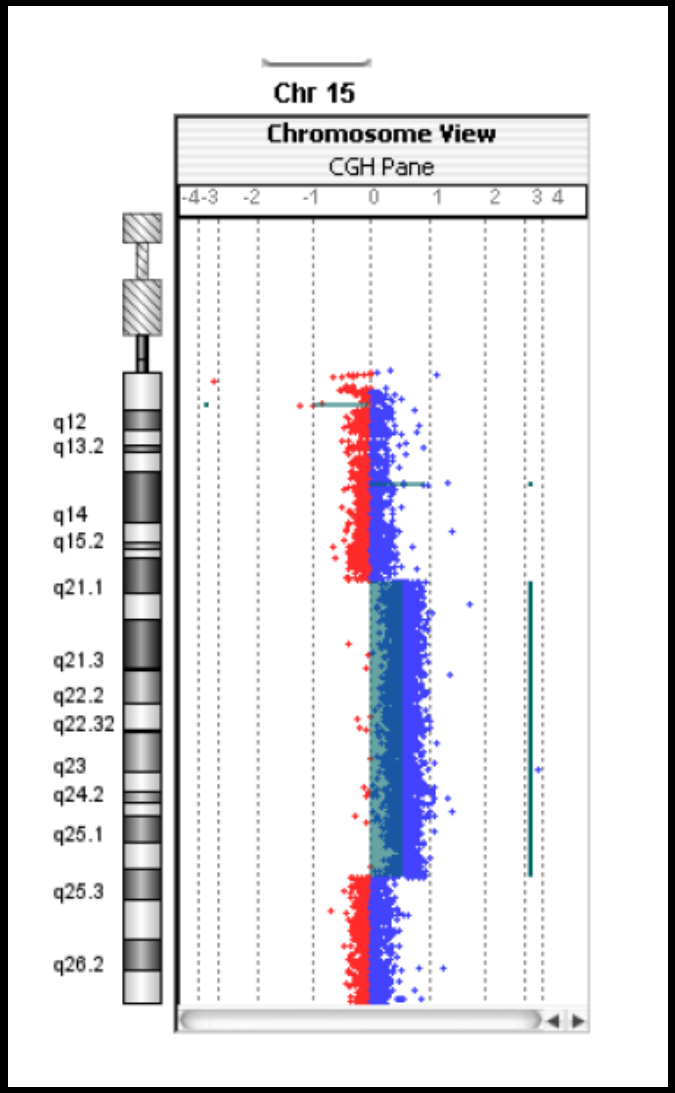
Клинический случай. Пациент П.



Клинический случай. Пациент П.



Клинический случай. Пациент П.



Ds: 46,XY,dup(15)(q21.1q25.3)

Клинический случай. Пациент П.

Amp/Del Intervals Table

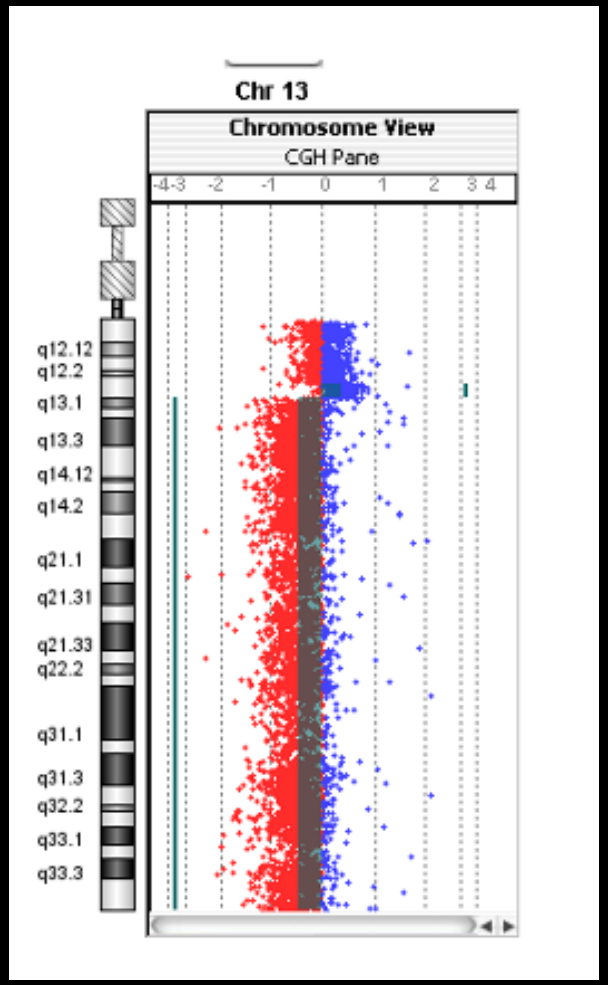
Chr	Start-Stop(bp)	Size(bp)	Cytoband	#Probes	Amp/Del	P-value	Annotations
chr1	196756231-196799302	43,072	q31.3	3	-0.927484	3.25E-11	CFHR3, CFHR1
chr2	242865920-243028452	162,533	q37.3	14	-0.905105	9.42E-46	
chr3	113573685-113614453	40,769	q13.31	3	0.897319	3.65E-11	GRAMD1C
chr3	162514534-162619141	104,608	q26.1	6	-0.913180	8.50E-22	
chr3	195425875-195634029	208,155	q29	21	0.306434	1.67E-11	MIR570, MUC20, MUC4...
chr4	69392545-69462438	69,894	q13.2	6	0.821321	6.14E-17	UGT2B17, UGT2B15
chr6	266079-362290	96,212	p25.3	8	-0.563730	1.03E-13	DUSP22
chr7	57253404-57558200	304,797	p11.2	12	-0.410054	2.52E-11	ZNF716
chr8	39237438-39374789	137,352	p11.22	12	0.878608	1.19E-37	ADAM5P, ADAM3A
chr10	46976157-47148546	172,390	q11.22	14	-0.746467	3.87E-38	GPRIN2, PPYR1, LOC728643
chr11	101306567-101412951	106,385	q22.1	8	0.527507	2.88E-12	TRPC6
chr14	20203449-20414232	210,784	q11.2	12	0.558845	5.16E-19	OR4Q3, OR4M1, OR4N2...
chr14	74001651-74022324	20,674	q24.3	3	0.825919	5.65E-11	HEATR4, ACOT1
chr15	24406896-24470140	63,245	q11.2	3	-0.995062	8.19E-14	PWRN2
chr15	34735949-34806953	71,005	q14	4	0.908696	6.03E-15	
chr15	47428535-86008048	38,579,514	q21.1 - q25.3	2824	0.539028	0.00E00	SEMA6D, SLC24A5, MYEF2...
chr16	32573808-33625989	1,052,182	p11.2	15	-0.479060	1.74E-17	TP53TG3B, TP53TG3, SLC6A10P
chr21	10866941-11000494	133,554	p11.2 - p11.1	7	0.499451	3.17E-10	TPTE
chr22	24347959-24407049	59,091	q11.23	6	-0.640220	3.26E-12	LOC391322, GSTT1, GSTTP2
chr22	39359112-39385485	26,374	q13.1	3	0.980345	1.02E-10	APOBEC3A, APOBEC3B

Amp=Amplification
Del=Deletion
Total Amp/Del Intervals: 20

Клинический случай. Пациент У.

Результат кариотипирования:
46,XY,-13,+mar

Клинический случай. Пациент У.

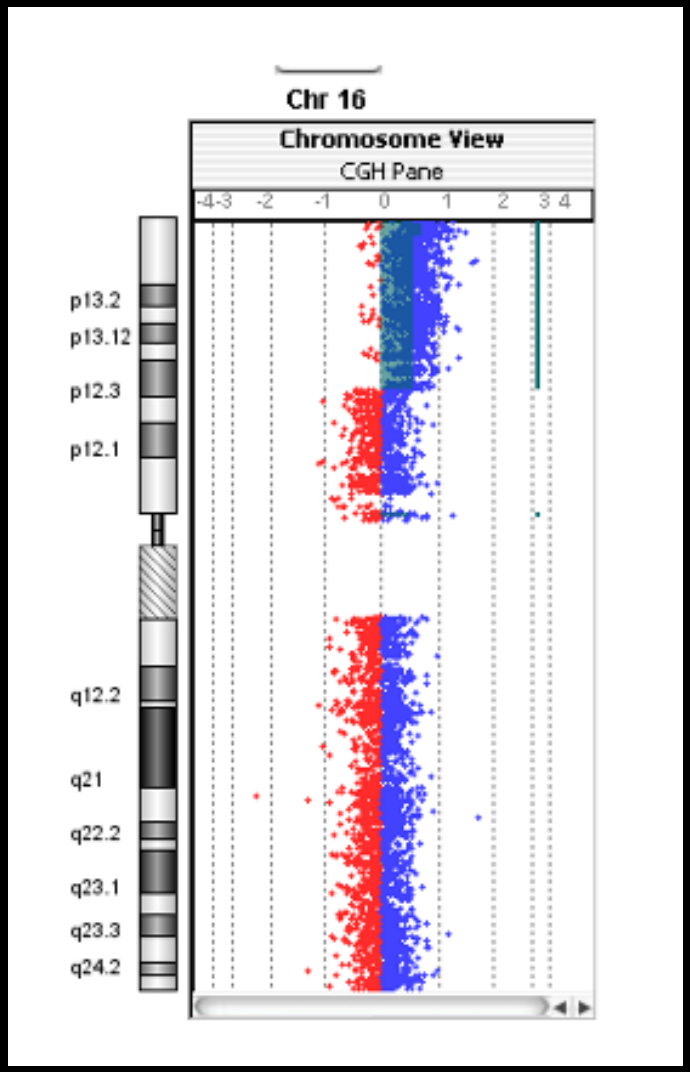


Уточненный диагноз:
46,XY,**der(13)del(q12.3~q34)**

Клинический случай. Пациент А.

Результат кариотипирования:
46,XY,der(7)

Клинический случай. Пациент А.



Ds:
46,XY,der(7)**t(7;16)(p22.1p12.3)**

Agilent CytoGenomics

Triage View : 1_252192418644 [Reviewed:SCANERPC\aekimov]

CytoGenomics

View | Copy | Search | Sample Info | QC Metrics | Change Call | Change Status | Reports

GenomeView(AMP: 0, DEL: 0, LOH: 0)

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y

ChromosomeView: chr1 (...

p36.21 p35.1 p32.2 p22.3 p13.3 q12 q21.3 q24.3 q31.2 q32.3 q42.2

-3 -2 -1 0 1 2 3

Gene View :chr1 : 0-1000000 , 1 Mb

100.0 Kb 200.0 Kb 300.0 Kb 400.0 Kb 500.0 Kb 600.0 Kb 700.0 Kb 800.0 Kb 900.0 Kb 1 Mb

Agilent Male CNV Re... | CNV-DGV_hg19_v3 | Pathogenic | Benign | MyDB | MyPatho

Probe Name	Chromosome	Start	Stop	Feature Number	Log Ratio
A_18_P10001390	chr1	759,762	759,821	60889	0.183
A_16_P15001594	chr1	852,863	852,922	15461	0.21
A_16_P00000136	chr1	890,945	891,004	49474	-0.175

Interval Table | CGH Probes | Single_Cell

RU 4:31 PM 9/9/2013

Agilent CytoGenomics

Triage View : Embryo_3_252192418647 [Reviewed:SCANERPC\aekimov]

CytoGenomics

View | Copy | Search | Sample Info | QC Metrics | Change Call | Change Status | Reports

GenomeView(AMP: 2, DEL: 2, LOH: 0)

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y

ChromosomeView: chrX (...

p22.2 p22.11 p11.4 p11.22 q11.2 q21.1 q21.32 q23 q25 q26.3 q28

-3 -2 -1 0 1 2 3

Gene View :chrX : 0-57464526 , 57.4 Mb

10.0 Mb 20.0 Mb 30.0 Mb 40.0 Mb 50.0 Mb

-3 -2 -1 0 1 2 3

Agilent Male CNV Re... | CNV-DGV_hg19_v3 | Pathogenic | Benign | MyDB | MyPatho

Chromosome	Start	Stop	Gene Name	Size(bp)	Type	#Probes	Mean Log Ratio	State	Suppress	Classification	Benign [81]	Actions
chrX SureFISH	2,318,311	54,562,094	ZBED1,CD99,XG,GYG	52,243,784	Amplification	926	0.575	Algorithm Generated	☐		1	Edit Notes
chrX SureFISH	63,006,748	154,290,166	FAM123B,MTMR8,M	91,283,419	Amplification	1,502	0.538	Algorithm Generated	☐		1	Edit Notes
chrY	2,656,461	9,940,478	RPS4Y1,ZFY,TGIF2LY	7,284,018	Deletion	55	-1.086	Algorithm Generated	☐		0	Edit Notes

Interval Table | CGH Probes | Single_Cell

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Agilent CytoGenomics

Triage View : 5G_252192418646_2_4 [Reviewed:SCANERPC\aekimov]

CytoGenomics

View | Copy | Search | Sample Info | QC Metrics | Change Call | Change Status | Reports

GenomeView(AMP: 0, DEL: 2, LOH: 0)

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y

ChromosomeView: chr2 (...

p24.3 p22.3 p16.2 p13.2 q13 q21.1 q23.2 q31.1 q32.2 q34 q37.1

-3 -2 -1 0 1 2 3

Gene View :chr2 : 102153997-137488953 , 35.3 Mb

105.0 Mb 110.00 Mb 114.99 Mb 120.0 Mb 125.0 Mb 130.0 Mb 135.0 Mb

Agilent Male CNV Re... | CNV-DGV_hg19_v3 | Pathogenic | Benign | MyDB | MyPatho

Probe Name	Chromosome	Start	Stop	Feature Number	Log Ratio
A_14_P130091	chr2	119,744,646	119,744,705	8334	0.136
A_14_P137818	chr2	119,833,027	119,833,086	22510	0.211
A_14_P123486	chr2	119,913,802	119,913,861	59492	-0.141

Interval Table | CGH Probes | Single_Cell

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Agilent CytoGenomics

Triage View : B7_252192418644 [Reviewed:SCANERPC\aekimov]

CytoGenomics

View | Copy | Search | Sample Info | QC Metrics | Change Call | Change Status | Reports

GenomeView(AMP: 19, DEL: 12, LOH: 0)

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y

ChromosomeView: chr1 (...

p36.21 p35.1 p32.2 p22.3 p13.3 q12 q21.3 q24.3 q31.2 q32.3 q42.2

-3 -2 -1 0 1 2 3

Gene View :chr1 : 0-127167224 , 127 Mb

25.0 Mb 50.0 Mb 75.0 Mb 100.0 Mb 125.0 Mb

Amp Amp Amp Amp

Agilent Male CNV Re... | CNV-DGV_hg19_v3 | Pathogenic | Benign | MyDB | MyPatho

Chromosome	Start	Stop	Gene Name	Size(bp)	Type	#Probes	Mean Log Ratio	State	Suppress	Classification	Benign [81]	Actions
chr1 SureFISH	1,054,735	120,529,725	MIR200B,MIR200A,M	119,474,991	Amplification	2,426	0.592	Algorithm Generated	☐		0	Edit Notes
chr1 SureFISH	144,988,715	249,138,851	SEC22B,HFE2,TXNIP,	104,150,137	Amplification	2,191	0.614	Algorithm Generated	☐		0	Edit Notes
chr2	1,112,915	9,327,795	TPO,PXDN,TSSC1,RN	8,214,881	Deletion	101	-0.649	Algorithm Generated	☐		0	Edit Notes

Interval Table | CGH Probes | Single_Cell

RU 4:29 PM 9/9/2013

Agilent CytoGenomics

Triage View : 1G_252192418646_1_4 [Reviewed:SCANERPC\aekimov]

CytoGenomics

View | Copy | Search | Sample Info | QC Metrics | Change Call | Change Status | Reports

GenomeView(AMP: 2, DEL: 20, LOH: 0)

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y

ChromosomeView: chr2 (...

p24.3 p22.3 p16.2 p13.2 q13 q21.1 q23.1 q31.1 q32.2 q34 q37.1

-3 -2 -1 0 1 2 3

Gene View :chr2 : 0-95010726 , 95.0 Mb

10.0 Mb 20.0 Mb 30.0 Mb 40.0 Mb 50.0 Mb 60.0 Mb 70.0 Mb 80.0 Mb 90.0 Mb

Agilent Male CNV Re... | CNV-DGV_hg19_v3 | Pathogenic | Benign | MyDB | MyPatho

Chromosome	Start	Stop	Gene Name	Size(bp)	Type	#Probes	Mean Log Ratio	State	Suppress	Classification	Benign [81]	Actions
chr2 SureFISH	42,444	90,012,396	ACP1,TPO,PXDN,TSS	89,969,953	Deletion	1,549	-0.589	Algorithm Generated	☐		0	Edit Notes
chr2 SureFISH	206,212	7,049,452	ACP1,TPO,PXDN,TSS	6,843,241	Deletion	89	-1.221	Algorithm Generated	☐		0	Edit Notes
chr2 SureFISH	96,074,013	243,041,364	ADRA2B,DUSP2,CIAO	146,967,352	Deletion	2,501	-0.527	Algorithm Generated	☐		0	Edit Notes

Interval Table | CGH Probes | Single_Cell

RU 4:23 PM 9/9/2013

Agilent CytoGenomics

Triage View : 3_2_252192418645 [Reviewed:SCANERPC\aekimov]

CytoGenomics

View | Copy | Search | Sample Info | QC Metrics | Change Call | Change Status | Reports

GenomeView(AMP: 4, DEL: 2, LOH: 0)

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y

ChromosomeView: chr5 (...

p15.2 p14.2 p13.1 q12.1 q13.3 q15 q22.1 q23.3 q32 q34 q35.3

-3 -2 -1 0 1 2 3

Gene View :chr5 : 17313921.75-77285827.25 , 59.9 Mb

30.0 Mb 40.0 Mb 50.0 Mb 60.0 Mb 70.0 Mb

Agilent Male CNV Re... | CNV-DGV_hg19_v3 | Pathogenic | Benign | MyDB | MyPatho

Chromosome	Start	Stop	Gene Name	Size(bp)	Type	#Probes	Mean Log Ratio	State	Suppress	Classification	Benign [81]	Actions
chr1 SureFISH	145,416,345	249,138,851	HFE2,TXNIP,RBM8A,	103,722,507	Amplification	2,109	0.413	Algorithm Generated	☐		0	Edit Notes
chr5 SureFISH	53,470,931	180,652,442	HSPB3,GZMA,CCNO,	127,181,512	Amplification	2,081	0.319	Algorithm Generated	☐		0	Edit Notes
chrX SureFISH	2,656,344	57,987,522	CD99,XG,GYG2,ARSD	55,331,179	Amplification	933	0.477	Algorithm Generated	☐		1	Edit Notes

Interval Table | CGH Probes | Single_Cell

RU 4:26 PM 9/9/2013

Точность и воспроизводимость

Multisample

Differential | Probe Penetrance | Interval Penetrance | Show | Copy | Go to | Setting | SNP | View In Table: Signal Intensity, Annotation

Chr 19

Chromosome View

CGH Pane

Chr 19: 29698345-30698345, 1 Mb

Custom Position

Gene View

CGH Pane

UQCRFS1, USTM2B, POP4, PLEKHF1, C19orf12, DCNE1, C19orf2

Tracks

MyDB

Cytoband_hg19_v2

29.6 Mb, 29.8 Mb, 30.0 Mb, 30.1 Mb, 30.3 Mb, 30.5 Mb, 30.6

CGH Probes

ProbeName	ChrName	Start	Stop	FeatureNum	567_252206027889_	468_252206022873_	468_252206022792_
A_16_P20986267	chr19	30,152,765	30,152,824	66523	0.174	0.270	0.235
A_14_P103669	chr19	30,162,231	30,162,289	156705	-0.019	0.017	0.136
A_14_P135370	chr19	30,173,190	30,173,249	36785	0.282	0.675	0.628
A_16_P03438010	chr19	30,184,067	30,184,126	167460	0.445	0.424	0.493
A_14_P133561	chr19	30,195,435	30,195,494	142097	0.587	0.675	0.592
A_14_P104329	chr19	30,203,903	30,203,962	70685	0.808	1.012	0.943
A_16_P41171718	chr19	30,216,967	30,217,026	110084	0.352	0.400	0.314
A_16_P03438088	chr19	30,242,167	30,242,226	150793	0.737	0.719	0.709
A_14_P128841	chr19	30,286,892	30,286,951	175962	0.522	0.818	0.341
A_14_P123495	chr19	30,304,361	30,304,420	24882	0.019	-0.108	-0.023

022060_20101001 | Selected Arrays

19:30198345 | Intra | ADM-2 | SN... | LOH | Default Featur... | Default Aberr... | Default Design... | G | C | D | F | hg19 | log2 ratio | Selected Row = 1931 | 4078 x 8

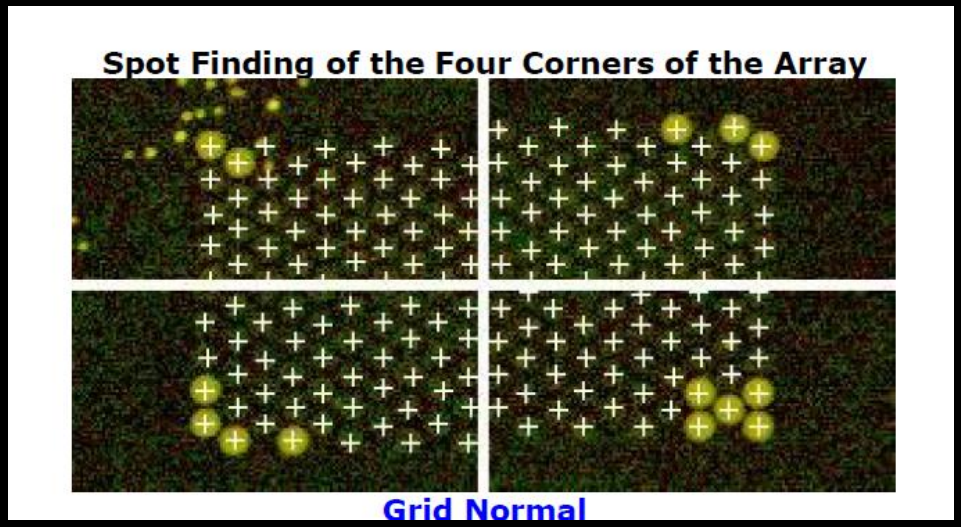
RU 8:35 AM 2/25/2014

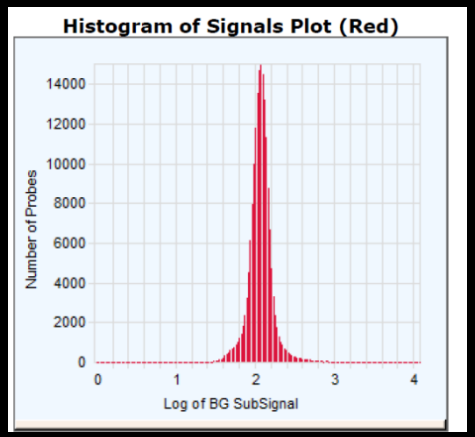
Контроль качества

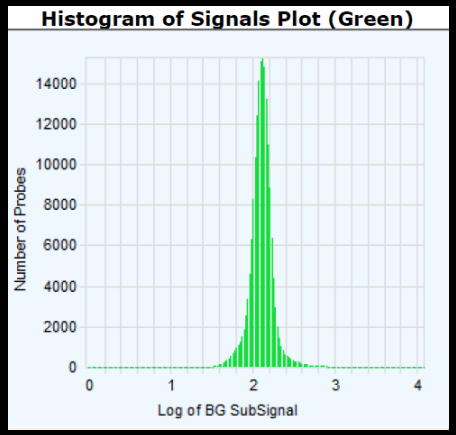
Evaluation Metrics for CytoCGH_QCMT_May11 :
Excellent (5) ; Good (6) ; Evaluate (1)

Metric Name	Value	Excellent	Good	Evaluate
IsGoodGrid	1.00	>1	NA	<1
AnyColorPrcntFeatNonUn...	0.00	<1	1 to 5	>5
DerivativeLR_Spread	0.18	<0.20	0.20 to 0.30	>0.30
gRepro	0.12	0 to 0.05	0.05 to 0.20	<0 or >0.20
g_BGNoise	3.61	<5	5 to 15	>15
g_Signal2Noise	31.43	>100	30 to 100	<30
g_SignalIntensity	113.52	>150	50 to 150	<50
rRepro	0.12	0 to 0.05	0.05 to 0.20	<0 or >0.20
r_BGNoise	3.28	<5	5 to 15	>15
r_Signal2Noise	31.61	>100	30 to 100	<30
r_SignalIntensity	103.52	>150	50 to 150	<50
RestrictionControl	-1.00		0.80 to 1	<0.80 or >1

♦ Excellent ♦ Good ♦ Evaluate







Клинический случай трисомия 21.

Клинический случай трисомия 18.

Синдром делеции 22q11.2

Синдром делеции 22q11.2

Синдром делеции 22q11.2

Синдром делеции 22q11.2

- Enter one or more search terms.
- Use **Limits** to restrict your search by search field, chromosome, and other criteria.
- Use **Index** to browse terms found in OMIM records.
- Use **History** to retrieve records from previous searches, or to combine searches.

NCBI is implementing changes to help you find current content in OMIM based on resources at NCBI, and then directing you to omim.org. Please be aware that you will leave NCBI to view OMIM records. Access to full records from NCBI (e.g. web, ftp, eutils) will no longer be supported.

OMIM ® - Online Mendelian Inheritance in Man ®

Welcome to OMIM ®, Online Mendelian Inheritance in Man ®. OMIM is a comprehensive, authoritative, and timely compendium of human genes and genetic phenotypes. The full-text, referenced overviews in OMIM contain information on all known mendelian disorders and over 12,000 genes. OMIM focuses on the relationship between phenotype and genotype. It is updated daily, and the entries contain copious links to other genetics resources.

This database was initiated in the early 1960s by Dr. Victor A. McKusick as a catalog of mendelian traits and disorders, entitled Mendelian Inheritance in Man (MIM). Twelve book editions of MIM were published between 1966 and 1998. The online version, OMIM, was created in 1985 by a collaboration between the National Library of Medicine and the William H. Welch Medical Library at Johns Hopkins. It was made generally available on the internet starting in 1987. In 1995, OMIM was developed for the World Wide Web by NCBI, the National Center for Biotechnology Information.

OMIM is authored and edited at the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, under the direction of Dr. Ada Hamosh.

NLM's Profiles in Science -- The McKusick Papers More...

NOTE: OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. While the OMIM database is open to the public, users seeking information about a personal medical or genetic condition are urged to consult with a qualified physician for diagnosis and for answers to personal questions.

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OMIM Entry - #235400 - HEM
Болезнь гемолитический уре
omim.org/entry/235400?search=cfhr&highlight=cfhr
Gmail - Входящие | Импортировано из IE | National Center for Bi... | SovMusic.ru - Вологда | Коллекция веб-фраг... | Коттеджный посело... | Рекомендуемые узлы

Table of Contents - #235400
External Links:
Clinical Resources
Animal Models
Cellular Pathways
Centers for Mendelian Genomics

#235400

HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 1; AHUS1

Alternative titles; symbols

AHUS, SUSCEPTIBILITY TO, 1

Other entities represented in this entry:

HEMOLYTIC UREMIC SYNDROME, TYPICAL, INCLUDED

Phenotype Gene Relationships

Location	Phenotype	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
1q31.3	{Hemolytic uremic syndrome, atypical, susceptibility to, 1}	235400	HF1	134370
1q31.3	{Hemolytic uremic syndrome, atypical, susceptibility to}	235400	CFHR3	605336
1q31.3	{Hemolytic uremic syndrome, atypical, susceptibility to}	235400	CFHR1	134371

Phenotypic Series

Clinical Synopsis

TEXT

A number sign (#) is used with this entry because susceptibility to the development of atypical hemolytic uremic syndrome-1 (AHUS1) can be conferred by variation in the gene encoding complement factor H (CFH; 134370) on chromosome 1q31 or by deficiency of the CFH-related proteins CFHR1 (134371) and CFHR3 (605366). Other genes may play a role in modifying the phenotype (see MOLECULAR GENETICS section).

HUS can share overlapping clinical features with thrombotic thrombocytopenic purpura (TTP; 274150), which is caused by mutation in the von Willebrand factor-cleaving protease (VWFCP) gene (ADAMTS13; 604134).

пуск | OMIM Entry - #2354... | Dropbox | Файлы | Microsoft PowerPoint ... | Microsoft Excel - Кни... | RL | 16:13

Inbox – aekimov@gmail | Скачанные файлы | decipher - Поиск в Google | DECIPHER v9.10: Mappin

Genome Research Limited [GB] | https://decipher.sanger.ac.uk

Сервисы | Bookmarks | USDRUB | Gmail | oparina4 | NCBI | Импортировано из | Дешевые авиабилет | Другие закладки

DECIPHER GRCh37

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DECIPHER is used by the clinical community to share and compare phenotypic and genotypic data. The DECIPHER database contains data from 21828 patients who have given consent for broad data-sharing; DECIPHER also supports more limited sharing via consortia. Have a look at the numbers.

Anyone can browse publicly-available patient data on DECIPHER and request to be put in contact with the responsible clinician. Why? Because sharing benefits everyone.

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As well as influencing individual patient outcomes, use of DECIPHER has contributed to over 1000 published articles since 2004. It's still free, and you are in control of what data to make public.

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Email

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Password

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ENG 18:28 12.10.2016

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https://www.clinicalgenome.org

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https://www.clinicalgenome.org/toolkits/

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Gene-Disease Clinical Validity Classifications
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Genomic Resources Search
Microarray Tracks
Pathogenicity Calculator
Preferred Condition List
Structural Variant Lab Contact Form
Structural Variation Database Search
VariantExplorer

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Microarray Tracks

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more »

Array Design Files For Array Analysis

Array design files for your cytogenomic microarray practice, including
design files and custom tracks to aid in analysis

more »

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Рекомендации по выбору метода исследования в зависимости от поставленной задачи.

Рекомендации по выбору метода исследования в зависимости от поставленной задачи.

- Природа нарушения (хромосомная, генная)
- Характер нарушения (инверсия, транслокация, мутация)
- Размер возможного нарушения (единичные замены и делеции, субмикроскопические измения, значительные делеции, дупликации)
- Исследуемый материал (кровь, хорион, амниоциты, абортивный материал)
- Сроки выполнения анализа (принципиально во время беременности)
- Техническая возможность применения
- Стоимость исследования.

Важность оформления сопроводительной документации для проведения анализа. Взаимодействие клиники и лаборатории.

Прошу провести данное исследование:	**Согласовано.** Заведующий лабораторией молекулярно генетических методов Трофимов Д.Ю.

Направление на анализ методом сравнительной геномной гибридизации (aCGH)

Фамилия:			
Имя:		Отчество:	
Возраст:		Пол:	
Наличие информированного согласия пациента:			
Биологический материал: кровь, хорион, бластомер, трофоэктодерма, другое			
Маркировка пробирки:			
Врач:	Фамилия		
	Подпись		
Отделение :			
Контактный телефон:			
Врач-генетик:	Фамилия		
	Подпись		
Предварительный диагноз:			
Результаты кариотипирования:			
Предполагаемое хромосомное нарушение (по возможности размер и позиция искомых CNV):			

Материал получил:

Число Подпись

- Интерпретация результатов проведения сравнительной геномной гибридизации в настоящее время является не тривиальной задачей

- Повышение разрешения метода по сравнению с традиционным кариотипированием приводит к экспоненциальному росту объема получаемой информации

- Количество получаемой в ходе анализа информации зачастую превосходит наши возможности по ее обработке

- Очень важным аспектом является разъяснение пациенту полученных результатов во избежание «самодиагностики»