

Unblocking sales by making the first ucer-centered DNA analysis tool in the undustry.

Singligitation

Context

iBinom is a DNA analysis SAAS platform that was providing mutation reports to doctors in US market. In 2015 team asked me to fix with design. With no idea on the field, I stepped in. It's an old case, but still one of the most complex ones I had in my life.

Chalenge We needed to fix the design in 4 weeks, right before the critical US conference. Design was blocking sales.

User experience

Doctors were reporting that platform is inconsistent, complex, and not optimised for laptop screens.

Scalability

Design architecture wasn't ready for the new databases, monetisation flows, and edge-cases.

Chr. Position	Ref	Δlt	Zugosity	Gene	rsID 1	Transcript ID	Amino acid d Quali	ity De	oth Effect V	ariant type Quality filter	Codon changlof	amd	nolyphen? tr	tationtast mutationa	ssefathmr	m sift	cadd	phylop46way p	hylon46v	(a) 1000gp1 af 100	10gn1 a	fr 1000gp1 e	u 1000gp1	am 1000gp1	asr esp6500	aa esn6500	ea HGMD	HGMD Phene clinyar	signif
100672	060 T	C	hom	DBT	rs12021720 E	ENST000003	p.Ser384Gly/	230	391 NON_SYNON SI	NP PASS	Agt/Ggt	iniu	0	1	1.2	T	-0.69	-0.21 1	.5	0.9 0.7	8 8	0.91	0.94	0.97	0.77	_aa_esposoo_ 0.9	no	Pathoge	enic
115231	254 G	A	het	AMPD1	rs61752479	ENST000005	p.Pro81Leu/c	220	447 NON_SYNON SI	NP PASS	cCg/cTg		1	0 1.4	-0.06	D	3.9	0.66 2	.8	0.054 0.0	061	0.11	0.086		0 0.023	0.13	yes	Adenosine m Pathoge	nic
115236	080 G	A	het	FMO3	rs17602729 E	ENST000003	p.Gin45*/c.1 p.Val257Met	210	40 NON_SYNON SI	NP PASS NP PASS	Gtg/Atg	1	0.0073	1 0.36	0.58	т	1.2	-1.2 -2	.1 2.2	0.094 0.0	28	0.058	0.083	0.2	0.024	0.13	yes	FMO3 varian Pathoge	anic
196659	237 C	Т	het	CFH	rs1061170 E	ENST000003	p.His402Tyr/	220	242 NON_SYNON SI	NP PASS	Cat/Tat		0.0005	1	-0.03	Т	-2.3	-4.4		-4 0.72 0.6	2	0.63	0.73	0.93	0.64	0.62	no	Pathoge	nic
203194 231408	186 C 091 A	G	het	GNPAT	rs11558492	ENST000003	p.Gly102Ser/ p.Asp519Gly	220	35 NON_SYNON SI	NP PASS NP PASS	Ggc/Agc gAt/gGt		0.031	1	3.8	D	1.8	4 0.66 0 0.46 -0	.32 0.13	0.3 0.2	9 75	0.32	0.29	0.3	0.27	0.29	no	Colorectal ca Pathoge Pathoge	anic
1 51549	496 T	С	het	MSMB	rs10993994	ENST000003	c88T>C	220	41 UPSTREAM SI	NP PASS						_											no	Pathoge	nic
1 70645	376 A 859 C	C T	het hom	STOX1 SAA1	rs10509305 E	ENST000002 ENST000003	p.Glu608Asp p.Ala70Val/c	220 190	35 NON_SYNON SI 31 NON_SYNON SI	NP PASS NP PASS	gaA/gaC gCc/gTc		0.006	1 -1.9	-0.81	3 T	-2.8	-2.5 -0	.003	0.14 0.0	28 5	0.21	0.24	0.096	0.058	0.21	no	Preeclampsic Pathoge Pathoge	nic enic
1. 122295	335 T	с	hom	HPD	rs1154510 E	ENST000002	p.Thr33Ala/c	230	55 NON_SYNON SI	NP PASS	Acg/Gcg			0	-0.38	Т	0.93	-0.23 1	.5	0.87 0.9	5	0.88	0.8	0.83	0.96	0.87	no	Pathoge	nic
1. 94847	415 A 053 G	G A	het het	SERPINA1 OCA2	rs6647 E	ENST000003 ENST000003	p.Val237Ala/ p.Arg305Trp,	220	41 NON_SYNON SI	NP PASS NP PASS	gTg/gCg Cgg/Tgg		0	1 -0.36 8 1.6	-1.8	D	0.65	-2.8 -2	.73	0.24 0.6	3	0.22	0.13	0.024	0.54	0.21	no yes	Darker eye ce Pathoge	nic enic
1 27374	180 T	с	het	IL4R	rs1805015 E	ENST000001	p.Ser503Pro,	220	34 NON_SYNON SI	NP PASS	Tcc/Ccc		0.12	1 1.1		3 T	2.1	0.46 0	.76	0.21 0.4	6	0.17	0.17	0.075	0.36	0.16	yes	Atopy, associ Pathoge	nic
1 56548	501 C 702 C	T T	hom het	BBS2 ASPA	rs4784677 E	ENST000002 ENST000002	p.Ser70Asn/c p.Tyr231Tyr/	230 220	322 NON_SYNON SI 314 SYNONYMOUSI	NP PASS NP PASS	aGc/aAc taC/taT			1	-1.2	T	0.72	-0.33 0	.95	1		1 0.99		1	1	1 0.99	no	Pathoge	nic enic
1 7125	591 T	c	het	ACADVL	rs113994167 E	ENST000005	p.Val306Ala/	220	377 NON_SYNON SI	NP PASS	gTg/gCg		0.39	1 0.95	-5.3	D	4.2	0.53		2					0.00023	0.0013	no	Pathoge	nic
1 42453 1 45360	065 A 730 T	c c	hom het	ITGA2B ITGB3	rs5911 E	ENST000002 ENST000005	p.lle874Ser/c p.Leu59Pro/c	230 220	22 NON_SYNON SI 434 NON_SYNON SI	NP PASS NP PASS	aTc/aGc cTg/cCg		0	0 -0.69	1.1	-3 T	0.49	-0.32		1 0.4 0.4 -4 0.092 0.1	3 2	0.4	0.36	0.42	0.39	0.37	yes yes	Reduced pos Pathoge Neonatal allc Pathoge	nic enic
2 46931	109 G	A	het	COL18A1	rs12483377 E	ENST000003	p.Asp1675As	220	515 NON_SYNON SI	NP PASS	Gac/Aac		1	0 1.6	0.6	т	4.2	0,66 2	.5	0.055 0.0	2	0.086	0.072	0.033	0.036	0.088	no	Pathoge	nic
2 18901	004 C 467 T	T G	hom het	PRODH MCCC1	rs450046 E	ENST000003 ENST000002	p.Arg521Gln, p.Arg385Ser,	230	33 NON_SYNON SI 468 NON_SYNON SI	NP PASS NP PASS	cGg/cAg agA/agC		0	0 1 4.8	1.6	-4 D	1.6 3.3	-0.47 0	.75 0.14	0.91 0.8		0.93	0.95	0.96	0.84	0.94	no yes	2-methylcrot Pathoge	anic
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10067206	кет				Zygosit	ty	Gene	1	SID			c Quai	117	Depth	201			variant typ		uality filte	er C	odon c	nang i	OT		nma		polypnenz_	јп n
11522125/			د ۸		hot				x61752470	ENSTODOOO	5 p.Ser 564Giy		250		147	NON_ST		SNP	P	455									<u>_</u>
11525125			^		het				~17602720	ENSTODOOO	p.FIOOILeu/	1	220		207			CNID		433	0				1		1		L
17100000			A ,		het		EMO2		\$17002729	ENSTODOOO	5 p.Gin45 /c	+	220		40	NON SVA			P/	455	C	ad/ 1aa			1		1	0.0072	
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19665923.	C		10	-	net		СЕН		\$1061170	ENSTODUOU	3 p.His402Tyr,	/	220	4	242 1	NON_SYN	NON	SNP	P,	ASS	0	at/lat	-				-	0.0005	+
203194186	5 C		T		het		CHIT1		s2297950	ENSTOOOOO	3 p.Gly102Ser	r/	220		79	NON_SYN	NON	SNP	P.	ASS	G	gc/Ago]	<u>L</u>
231408093	A		G		het		GNPAT	!	s11558492	ENST00000	3 p.Asp519Gly	V	220		35	NON_SYN	NON	SNP	P,	ASS	g	At/gGt						0.031	_
51549496	5 T		С		het		MSMB	!	s10993994	ENST00000	3 c88T>C	-	220		41	UPSTREA	M	SNP	P	ASS	-								_
70645376	5 A		с		het		STOX1		s10509305	ENST00000	2 p.Glu608As	р	220		35	NON_SYN	NON	SNP	P,	ASS	g	aA/gaC)
18290859	9 C		T		hom		SAA1		s1136743	ENST00000	3 p.Ala70Val/0	c	190		31	NON_SYN	NON	SNP	P,	ASS	g	Cc/gTc						0.006	_
122295335	5 T		с		hom		HPD	!	s1154510	ENST00000	2 p.Thr33Ala/	c	230		55	NON_SYN	NON	SNP	P.	ASS	A	cg/Gcŧ	3						-
94847419	5 A		G		het		SERPINA	.1 I	s6647	ENST00000	3 p.Val237Ala,	/	100		39	NON_SYM	NON	SNP	P,	ASS	g	Tg/gCg						()
28260053	3 G		A		het		OCA2		s1800401	ENST00000	3 p.Arg305Trp),	220		41	NON_SYM	NON	SNP	P.	ASS	C	gg/Tgg						0.42	0
27374180	т		с		het		IL4R	1	s1805015	ENST00000	1 p.Ser503Pro)/	220		34	NON_SYN	NON	SNP	P,	ASS	To	cc/Ccc						0.12	
56548503	ιc		Т		hom		BBS2	j	s4784677	ENST00000	2 p.Ser70Asn/	/<	230		322	NON_SYN	NON	SNP	P,	ASS	a	Gc/aAc	:						
3397702	2 C		Т		het		ASPA	1	s12948217	ENST00000	2 p.Tyr231Tyr,	1	220	3	314	SYNONY	νοι	SNP	P,	ASS	ta	C/taT							
7125593	T		С		het		ACADVL	1	s113994167	ENST00000	5 p.Val306Ala,	/	220	3	377	NON_SYN	NON	SNP	P	ASS	g	Tg/gCg						0.39	
42453065	5 A		С		hom		ITGA2B	1	s5911	ENST00000	2 p.Ile874Ser/	/c	230		22	NON_SYM	NON	SNP	P.	ASS	a	Tc/aGc						()
45360730	Т		с		het		ITGB3	1	rs5918	ENST00000	5 p.Leu59Pro/	/0	220	4	434	NON_SYM	NON	SNP	P.	ASS	c	∏g/cCg						0.005	
46931109	G		A		het		COL18A	1 I	s12483377	ENST00000	3 p.Asp1675A	s	220	Ç	515	NON_SYM	NON	SNP	P.	ASS	G	ac/Aac							L
18901004	t C		т		hom		PRODH	1	s450046	ENST00000	3 p.Arg521Glr	n,	230		33	NON_SYM	NON	SNP	P	ASS	c	Gg/cAg	3					()
18275946	T		G		het		MCCC1	1	s119103213	ENST00000	2 p.Arg385Ser	r/	220	4	468	NON_SYN	NON	SNP	P.	ASS	a	gA/agC						0.88	

Printed report. It was impossible to print 40 columns of data on A4.





Service agreement About Manual

Raw data (43.22 Gb of 250.00 Gb) Results

Select some files for example and you will see how it works



Sort by name

Homescreen & upload. Unclear hierarchy, navigation and layout.



ignat1990@gmail.com (Sign out) 0 panels, 16 exomes, 0 genomes



Press to upload or drag'n'drop

Analyze

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Raw data **Results**

You can download result files here

Move to archive Solution Delete	
Sample Aligned reads (bam + bai) VCF Filter	Report
Archive (16 files)	
L001_I002_KJSC120914KJ216B.bam	Filter
sample.fastq.gz (7)	Filter
sample.fastq.gz (8)	Filter
sample.fastq.gz (9)	
sample.fastq.gz (10)	<u>Filter</u>
Myodystrophy_M	Filter
short.fastq.gz (1)	Filter
001.fastq	Filter
sample.fastq.gz (6)	Filter
Pomykalov	Filter
Zernov 018 deafness	<u>Filter</u>
Zernov 002 hypotrihosis	<u>Filter</u>
	 Move to archive Sample Aligned reads (bam + bai) VCF Filter name ▲ Sample Aligned reads (bam + bai) VCF Filter Archive (16 files) L001_1002_KJSC120914KJ216B.bam sample.fastq.gz (7) sample.fastq.gz (8) sample.fastq.gz (8) sample.fastq.gz (9) sample.fastq.gz (10) Myodystrophy_M short.fastq.gz (1) 001.fastq sample.fastq.gz (6) Pomykalov Zernov 018 deafness Zernov 002 hypotrihosis

Processed results. Unclear, inconsistent and complex structure.

Varia	ant call (vcf)	Variant call (csv)	Date
<u>3.48 Mb</u> 😔	25.51 Mb 😔	<u>14.40 Mb</u> 😔	07.04.2015 03:49:53
<u>745.47 Kb</u> ⊙	<u>19.85 Mb</u> 😔	<u>8.82 Mb</u> 😔	07.05.2015 02:01:20
<u>745.46 Kb</u> ⊙	<u>19.85 Mb</u> 😔	<u>8.82 Mb</u> 😔	08.05.2015 20:40:08
<u>718.35 Kb</u> ⊙	<u>0 bytes</u> 😔	<u>0 bytes</u> 📀	13.05.2015 13:56:09
<u>744.97 Kb</u> 📀	20.01 Mb 😔	<u>8.82 Mb</u> 😔	13.05.2015 20:54:12
<u>745.35 Kb</u> 😔	<u>42.46 Mb</u> 😔	<u>19.51 Mb</u> 😔	29.05.2015 19:50:09
<u>746.90 Kb</u> ⊙	<u>33.42 Kb</u> 😔	<u>12.95 Kb</u> 😔	02.06.2015 16:00:26
<u>744.88 Kb</u> ⊙	<u>90.87 Kb</u> 😔	<u>41.42 Kb</u> 😔	04.06.2015 13:43:53
<u>745.46 Kb</u> ⊙	<u>19.85 Mb</u> 😔	<u>8.82 Mb</u> 😔	06.05.2015 14:34:09
<u>745.41 Kb</u> 😔	<u>17.36 Mb</u> 😔	<u>8.29 Mb</u> 😔	09.06.2015 18:38:51
<u>745.14 Kb</u> ⊙	<u>33.16 Mb</u> 😔	<u>15.49 Mb</u> 😔	09.06.2015 23:06:10
<u>745.10 Kb</u> ⊙	<u>31.27 Mb</u> 😔	<u>14.57 Mb</u> 😔	14.06.2015 01:48:50



<i>i</i> BINOM Sample r	name: /L001_I002_KJSC120914KJ216B.bam			
Input Data 101461				
funclass Codon change	Distance to transcript Amino acid change	Amino acid length Gene lof_perc nmd_perc	polyphen2_hvar_score mutationtaster_score	
		TUBB8P11 TUBB8P11		
	208	18 AL645608.2		
B II	NOM Sample	name: /L001_l002_KJSC	120914KJ216B.bam	
Input Da 10146	ata			
funclass	Codon change	Distance to transcript	Amino acid change	Amino acid lengt
		208		18

Web report. Bad usability-table was 3 times wider than regular screen.

_						
5	Gene	lof_perc	nmd_perc	polyphen2_hvar_score	mutationtaster_score	mutationasses
	TUBB8P11					
	TUBB8P11					
	AL645608.2					

	EXON		
	EXON		
	UPSTREAM		

⊕ Q
sor_score

Process

To fit in 4 weeks, I had four processes at the same time. Each of them was giving valuable feedback.

Expert interviews Learning the field and user specifics. Testing the design. Reports design lterating on the optimal datavisualisation and look. Web design Applying new ideas from the reports. Drafting the architecture. Frontenddevelopment Delivering the ready parts and reviewing the prototypes.

Mutations		
ADF: Non synonymous coding Chr-1, position-123123, hom (ENST 0000037012)	Clinvar: patogenic HKMD: adenosine, monophos,hate, deaminase, deficiency	ACMG-7
p.Ser384Gly/c.1150A>G G→A RS12417413	L 0,5 0,8 L 0,5 0,8 Taster 0,2 0,03 0,05 0,8 1 0,03 0,05 0,05	0,02 0,02 0,02 amr asn 0,02 eaaf eaaf esp600
ADF: Non synonymous coding Chr-1, position-123123, hom (ENST 0000037012)	Clinvar: patogenic HKMD: adenosine, monophos,hate, deaminase, deficiency	ACMG-7
p.Ser384Gly/c.1150A>G G→A RS12417413	L 0,5 0,8 L 0,5 0,8 0,1 0,1 primate Pacential taster 0,2 0,03 0,05 0,1 0,1 primate Pacential taster 0,2 0,03 afr 0,03 average afr 0,03 average afr 0,03 eur eur eur gvar	0,02 0,02 0,02 amr asn aaaf 0,02 esp600
ADF: Non synonymous coding Chr-1, position-123123, hom (ENST 0000037012)	Clinvar: patogenic HKMD: adenosine, monophos,hate, deaminase, deficiency	ACMG-7
p.Ser384Gly/c.1150A>G G→A RS12417413	L 0.5 0.8 L 0.3 0.5 0.8 0.1 0.3 primate Pacential Lof Nmd Poly Fath Sift Cadd Phylop 46way Mutation 1000gp1 a gvar	0,02 0,02 0,02 amr asr aaaf 0,02 esp600

Mutations

Gene and effect ADF homozygote non synonymous	Status Clinvar: patogenic	Acid change p.Ser384Gly/c.1150A>G	acmg 7
coding Chr-1, position-123123 (ENST 0000037012)	HKMD: adenosine, monophos, hate, deaminase, deficiency	G→A RS12417413	
Lof Nmd Polyphen2 Fathmm Sift Cadd Phyl	op 46way Mutation 1000)gpla	esp600

Lof	Nmd	Polyphen2 gvar	Fathmm	Sift	Cadd	Phylop primate	46way pacentai	Mutat taster	ion asessor	1000gpl average	a afr	eur	amr	asn	esp600 aaaf	eaaf
I	0, I	0,5	0,8		0, I	0,3	0,5	0,8	0,2	0,03	0,05	0,03	0,02	0,02	0,02	0,02

Printed report. It took 2 weeks and 30 iterations to fit 40 columns of data in one component with clear logic and hierarchy.

WASH7P: EXON	Scores Clinvar-, ACMG-7
n.1493G>A	fathmm mutation taster primate phylop46 poly phen2 mutation assessor placental phylop46 cadd
no:	Frequency 0,01–0,0001
Chr-1, position-14653, het	1000GPI ESP6500
ENST 00000438504, RS 375086259	avr afr eur amr asn ea aa
DDXIILI: DOWNSTREAM	Scores Clinvar-, ACMG-7
n.*1657A>G	fathmm mutation taster primate phylop46 poly phen2 mutation assessor placental phylop46 cadd
no:	Frequency 0,01–0,0001
Chr-1, position-14907, hom	1000GP1 - ESP6500
ENST 00000456328, RS79585140	avr afr eur amr asn ea aa
DDXIILI: DOWNSTREAM	Scores Clinvar-, ACMG-7
n.*1657A>G	fathmm mutation taster primate phylop46 poly phen2 mutation assessor placental phylop46 cadd
no:	Frequency 0,01–0,0001
Chr-1, position-14930, hom	1000GPI ESP6500
ENST 00000456328, RS75454623	avr afr eur amr asn ea aa
DDXIILI: DOWNSTREAM	Scores Clinvar-, ACMG-7
n.*1657A>G	fathmm mutation taster primate phylop46 poly phen2 mutation assessor placental phylop46 cadd
Frequency (0, 1–0,0001) 1000GPI avr 0,05 afr 0,01 eur 0,11 amr 0,09 asn ESP6500 aa 0,09 ea 0,00	0,00 -0,06 0,0 0,66 1,0 1,4 2,8 3,9 fathnm mutation primate phylop46 phen2 asessor phylop46 phylop46
ADF: Non synonymous coding Patogenic A	denosine, monophos, hate, deaminase, deficiency
Chr-1, position-123123, homozygote Clinvar HI	KMD

ADF: Non synonymous coding Chr-1, position-123123, homozygote						Pa Clii	ntogenic	genic Adenosine, monophos, hate, deaminase, нкмр						deficiency RS124				
F	requency 000GP1	0,05 avr	0–0,0 0,01 afr	001) 0,11 eur	0,09 amr	0,000 I asn	ESP6500	0,00 ea	0,09 aa	Scores -0,06 fathnm	6 (ACMG 0,0 mutation taster	0,66 primate phylop46	I,0 poly phen2	I,4 mutation asessor	2,8 placental phylop46	3,9 cadd	Amino-acid change p.Ser384Gly/c.1150A>G (G→A)	



Readability. To ensure readability of a report in real life, we were printing each new version to hold it in hands.





Status Acid change Clinvar: patogenic p.Ser384Gly HKMD: adenosine, monophos, GA RS1241 hate, deaminase, deficiency	La NULERIA Narithe Barry Narithe Barry North Contention North Contention N	And the second s	Book Star. Book Star. Book Star. Star. Star. I I I I I I I I I I I I I I I I I I I I	N Keedbard2000	VCF Filter	PCS d to report nymous coding ENS G, (G→A), Ch∩1, por-115231254, homozygote nophosp, hate, deaminase, deficency		errbit.ibinom.com	ADF: Non synonymous coding p.Ser384Gyic IIS0A>G. (G→A) HGMD:Adenoune, morophos, hate, deaminase, deficency Chr-1 pop-123123/homozygote ENST 0000037012. RS12417413 -: {{ Trimstrine(filterservice.data[idx]		Search
Phylop 46way Mutation 1000gp1 a primate pacentai taster asessor average afr eu 0,3 0,5 0,8 0,2 0,03 0,05 0,	Annum Verpa C. Ann. Annum Verpa C. Ann. Marchan Strategy (March 1996) (March 1996) (March 1 Marchan March 1996) (March	Anaroma an	The second secon	NUTTION Ans. Soc. $S \in S \in S$		enic,ACMG-7 0,66 I,0 I,4 2,8 primatephylop16 polyphen2 mutationasessor placentalphylop1 I 0.05 0.01 0,11 0.09 0,0001 ESP6500 0,00 0,09 avr afr eur ann asn ea aa	part IBUDS Assessment PARM Mich. V. Nord, D.M. 200000000000000000000000000000000000		<pre>[filterservice.headersnames.indexof('effect')]) }} .(-) HGMD:- Chr0.position</pre>		acient
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deversion yours and party and party in the your state your your your your and party and you	Aussidiardigues KCP APOL APO FAIler (1921-1) 1921 (1926 (1.506 ()) Bud scannel (COLORIS) FRancisca Annual () Francisca Annual () Rear Call () area () () Rear Call ()	Anomardigues (137 1903 AN FLAT FLAT FLAT (138)) (109)) Reference States Photoge States The Association (139) The Association (139) T) 9 0 14 0 19 0) 10 0 15 0 20 0	AD. An approprint (PA) Model (PA) Model (PA) AD. An approprint (PA) Model (PA) <th>State State State</th> <th></th> <th>Come BO Edition Boot 2000 Frequency 0.9–0.0001 1000GPI 0.05 0.01 0.09 0.0001 ESP6500 0.00 0.09 1000GPI 0.05 0.01 0.09 0.0001 ESP6500 0.00 0.09 1000GPI 0.05 0.01 0.01 0.09 0.0001 ESP6500 0.00 0.09</th> <th>i alaysis in 2016. By that t</th> <th>cients genome scan to create</th> <th>create new pacient</th> <th></th>	State		Come BO Edition Boot 2000 Frequency 0.9–0.0001 1000GPI 0.05 0.01 0.09 0.0001 ESP6500 0.00 0.09 1000GPI 0.05 0.01 0.09 0.0001 ESP6500 0.00 0.09 1000GPI 0.05 0.01 0.01 0.09 0.0001 ESP6500 0.00 0.09	i alaysis in 2016. By that t	cients genome scan to create	create new pacient	
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			Sc 0,09 -0, aa fat	,00 -0,06 0,0 fathnm mut taste	0000 afr 0,01 eur 0,1	Taguer 1994, mail entia, progressing tations uman genes are deciphered, so it is possible that the seat does int revea foull need to pass the another alaysis in 2016. By that time sciencitists mother 10% PARR PERN 1. PERN 1. DOM: 1 State science genes EGV 137L UPO EPX S	tHandin.27 august 1994.main Demonstration progressing mutations d consultation docton-genetics. Because of science progress.new is will be discovered. Need to make another analysis in 2016. Sated genes MOE.APT. PRARE MEN. L. PSEN 1, 10AL 1		30 pload	002_1005_KJSC120914KJ217 3.0 002_1006_KJSC120914KJ217B 3.1 002_1007_KJSC120914KJ217 3.0 002_1008_KJSC120914KJ217 3.0	Genome scans Drag scan here to upload test.fastq (2) ham 31 Ho L001_1004_KJSC120914KJ21 L002_1008_KJSC120914KJ21
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	Fair Fail (20 ⁻ 12)(20 ⁺ 12) ⁻¹² (20 ⁺ 12) ⁻¹² Papelolos Fuguero (Inde) + 15	Transform Transform (SUN) (171)		0		n-881627,	Marked Street St	CONTRACTOR CONTRACTON CONTRACTON CONTRACTON CONTRACTON CONTRACTON C		Constitution (Constitution) Constitution (Co	<u>3.46 Mb</u> ⊙ <u>3.46 Mb</u> ⊙

Process. Alignment was critical, so each day we had a screenshot ping-pong with devs.





Printed report. We found structure that was both readable, logic, and

scalable. Team confirmed it with user testing.

Web application.

The new version was delivered in time. To fit in the deadline, we cut out some of the features and sacrificed refactoring.

Instrument: Illumina HiSeq Library: mate-pair Specimen: whole blood Bad scanned genes!

Database version ClinVar 1.2, apr'15, CADD 1.0, may'14

PolyPhen 1.2, dec'14 ExAc 1.3, aug'13 HGMD 0.31, sept'14 1000Genomes 0.3, jan'13



Dementia, progressing. Mutations report

Robert Hairullin, 27 august 1994, male

Filter: DP>20, QUAL>20, Missense, Population frequency ExAc<1%.

o.Ser384Gly/c.1150A>G (gaC/gaT)	4	Mutationtaster	0,0 (46)	1000GPI	ESP6500	ExAc
HGMD Adenosine monophosp hate deaminase deficency	ACMG	Mutationassessor	1,4 (30)	Avr 0,05	Ea 0,05	Avr 0.04
Clinvar Patogenic		Placentalphylop46	0.66 (21)	Afr 0,01	Aa 0,01	
		Primatephylop46	2.8 (15)	Eur 0,11		
NOTES ENST	0000037012	Polyphen2	1,0 (32)	Amr 0,09		
R	\$ 12417413	Cadd	3,9 (11)	Asn 0,0001		
P	os 1:321313	Lof	-0,06 (5)			
		Nmd	-			
		Fathnm	-			
ADF. Homozygote non synonymous coding (SNP)	1	Scores		Frequency		
p.Ser384Gly/c.1150A>G (gaC/gaT)	4	Mutationtaster	0,0 (46)	1000GPI	ESP6500	ExAc
HCMD. Adenosing monophose bate desminase deficency	ACMG	Mutationassessor	1,4 (30)	Avr 0,05	Ea 0,05	Avr 0,04
Clinvar Patogenic		Placentalphylop46	0,66 (21)	Afr 0,01	Aa 0,01	
		Primatephylop46	2,8 (15)	Eur 0,11		
		Polyphen2	1.0 (32)	Amr 0,09		
NOTES ENST	0000037012 \$12417413	Cadd	3,9 (11)	Asn 0,0001		
P	os 1:321313	Lof	-0,06 (5)			
		Nmd	-			
		Fathnm	2			
		1 80019362.02				
	-	Connection		E		
ADF. Homozygote non synonymous coding (SNP)	5	Scores	00/10	Frequency	ECD/FAA	EuAa
p.Ser384Gly/c.1150A>G (gaC/gaT)	5	Mutationtaster	0,0 (46)	TOUGPT	ESP6500	EXAC
HGMD Adenosine, monophosp, hate, deaminase, deficency.	ACMG	Mutationassessor	1,4 (30)	Avr 0.05	Ea 0.05	Avr 0.04
Clinvar Patogenic		Placentalphylop46	0,66 (21)	Afr 0,01	Aa 0,01	
		Primatephylop46	2,8 (15)	Eur 0,11		
NOTES ENST	0000037012	Polyphen2	1.0 (32)	Amr 0,09		
R	\$ 12417413	Cadd	3,9 (11)	Asn 0,0001		
P	os 1:321313	Lof	-0.06 (5)			
		Nmd				
		T SING				
		Fathnm	-			
		Fathom	-			
ADF. Homozygote non synonymous coding (SNP)	2	Fathnm Scores	-	Frequency		
ADF. Homozygote non synonymous coding (SNP) 5.5er384Gly/c.1150A>G (gaC/gaT)	3	Fathnm Scores Mutationtaster	0.0 (46)	Frequency 1000GP1	ESP6500	ExAc
ADF. Homozygote non synonymous coding (SNP) p.Ser384Gly/c.1150A>G (gaC/gaT)	3 Acmg	Fathnm Scores Mutationtaster Mutationassessor	- 0,0 (46) 1,4 (30)	Frequency 1000GPI Avr 0,05	ESP6500 Ea 0.05	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp. hate, deaminase, deficency.	З	Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46	0.0 (46) 1.4 (30) 0.66 (21)	Frequency 1000GPI Avr 0,05 Afr 0,01	ESP6500 Ea 0,05 Aa 0,01	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp. hate, deaminase, deficency. Clinvar Patogenic	З	Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46	0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15)	Frequency 1000GP1 Avr 0,05 Afr 0,01 Eur 0,11	ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) p.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp. hate, deaminase, deficency. Clinvar Patogenic	З	Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46 Polyphen2	- 0,0 (46) 1,4 (30) 0,66 (21) 2,8 (15) 1,0 (32)	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09	ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp. hate, deaminase, deficency. Clinvar Patogenic NOTES ENST	3 ACMG	Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46 Polyphen2 Cadd	0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32) 3.9 (11)	Frequency 1000GP1 Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001	ESP6500 Ea 0,05 Aa 0,01	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp. hate, deaminase, deficency. Clinvar Patogenic NOTES ENSTO R	3 ACMG 0000037012 5 12417413 os 1:321313	Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46 Polyphen2 Cadd Lof	0,0 (46) 1,4 (30) 0,66 (21) 2,8 (15) 1,0 (32) 3,9 (11) -0,06 (5)	Frequency 1000GP1 Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001	ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) p.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp. hate, deaminase, deficency. Clinvar Patogenic NOTES ENSTO R Pi	3 ACMG 0000037012 5 12417413 os 1:321313	Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46 Polyphen2 Cadd Lof Nmd	0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32) 3.9 (11) -0.06 (5)	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001	ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) p.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp. hate, deaminase, deficency. Clinvar Patogenic NOTES ENST(R Pi	3 ACMG 0000037012 (\$ 12417413 os 1:321313	Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46 Polyphen2 Cadd Lof Nmd Fathnm	0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32) 3.9 (11) -0.06 (5)	Frequency 1000GP1 Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001	ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp. hate, deaminase, deficency. Clinvar Patogenic NOTES ENSTO R Pi	3 ACMG 0000037012 5 12417413 os 1:321313	Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46 Polyphen2 Cadd Lof Nmd Fathnm	0,0 (46) 1,4 (30) 0,66 (21) 2,8 (15) 1,0 (32) 3,9 (11) -0,06 (5) -	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001	ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ENST(R Pi	3 ACMG	Fathnm Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46 Polyphen2 Cadd Lof Nmd Fathnm	0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32) 3.9 (11) -0.06 (5) -	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001	ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ENST(R PI ADF. Homozygote non synonymous coding (SNP)	3 ACMG 0000037012 (\$ 12417413 05 1:321313	Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46 Polyphen2 Cadd Lof Nmd Fathnm Scores	- 0,0 (46) 1,4 (30) 0,66 (21) 2,8 (15) 1,0 (32) 3,9 (11) -0,06 (5) - -	Frequency 1000GP1 Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001	ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0,04
ADF. Homozygote non synonymous coding (SNP) D.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp. hate, deaminase, deficency. Clinvar Patogenic NOTES ENST(R Pi ADF. Homozygote non synonymous coding (SNP) D.Ser384Gly/c.1150A>G (gaC/gaT)	3 ACMG	Fathnm Scores Mutationtaster Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46 Polyphen2 Cadd Lof Nmd Fathnm Scores Mutationtaster	0,0 (46) 1,4 (30) 0,66 (21) 2,8 (15) 1,0 (32) 3,9 (11) -0,06 (5) - - - 0,0 (46)	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001 Frequency 1000GPI	ESP6500 Ea 0.05 Aa 0.01 ESP6500	ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ENSTO R P ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD. Adenosine, monophosp hate, deaminase, deficency	3 ACMG 0000037012 5 12417413 bs 1:321313	Fathnm Fathnm Scores Mutationtaster Mutationtaster Placentalphylop46 Primatephylop46 Polyphen2 Cadd Lof Nmd Fathnm Scores Mutationtaster Mutationtaster Mutationassessor	0,0 (46) 1,4 (30) 0,66 (21) 2,8 (15) 1,0 (32) 3,9 (11) -0,06 (5) - - - - 0,0 (46) 1,4 (30)	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001 Frequency 1000GPI Avr 0,05	ESP6500 Ea 0.05 Aa 0.01 ESP6500 Ea 0.05	ExAc Avr 0.04 ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ENST(R P ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic	3 ACMG 0000037012 (\$ 12417413 005 1:321313	Fathnm Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Polyphen2 Cadd Lof Nmd Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46	0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32) 3.9 (11) -0.06 (5) - - 0.0 (46) 1.4 (30) 0.66 (21)	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001 Frequency 1000GPI Avr 0,05 Afr 0,01	ESP6500 Ea 0.05 Aa 0.01 ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0,04 ExAc Avr 0,04
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ADF. Homozygote non synonymous coding (SNP) o.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ENST(R P ADF. Homozygote non synonymous coding (SNP) o.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic	3 ACMG 0000037012 5 12417413 os 1:321313	Fathnm Fathnm Scores Mutationtaster Mutationtaster Placentalphylop46 Primatephylop46 Polyphen2 Cadd Lof Nmd Fathnm Scores Mutationtaster Mutationtaster Mutationtaster Placentalphylop46 Primatephylop46 Polyphen2	0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32) 3.9 (11) -0.06 (5) - - 0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32)	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001 Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09	ESP6500 Ea 0.05 Aa 0.01 ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0.04 ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) a.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ENST(R P ADF. Homozygote non synonymous coding (SNP) a.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ENST(R	3 ACMG 0000037012 5 12417413 05 1:321313 ACMG 0000037012 5 12417413	Fathnm Fathnm Scores Mutationtaster Mutationtaster Mutationassessor Placentalphylop46 Polyphen2 Cadd Lof Nmd Fathnm Scores Mutationtaster Mutationtaster Mutationtaster Placentalphylop46 Primatephylop46 Polyphen2 Cadd	0,0 (46) 1,4 (30) 0,66 (21) 2,8 (15) 1,0 (32) 3,9 (11) -0,06 (5) - - 0,0 (46) 1,4 (30) 0,66 (21) 2,8 (15) 1,0 (32) 3,9 (11)	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001 Frequency 1000GP1 Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001	ESP6500 Ea 0.05 Aa 0.01 ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0.04 ExAc Avr 0.04
ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ADF. Homozygote non synonymous coding (SNP) b.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ENST(R P	3 ACMG 0000037012 5 12417413 05 1:321313 ACMG 0000037012 5 12417413 05 1:321313	Fathnm Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Polyphen2 Cadd Lof Nmd Fathnm Scores Mutationtaster Mutationtaster Mutationassessor Placentalphylop46 Primatephylop46 Polyphen2 Cadd Lof Cadd Lof	0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32) 3.9 (11) -0.06 (5) - - 0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32) 3.9 (11) -0.06 (5)	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001 Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001	ESP6500 Ea 0.05 Aa 0.01 ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0,04 ExAc Avr 0,04
ADF. Homozygote non synonymous coding (SNP) a.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ENSTO R PADF. Homozygote non synonymous coding (SNP) a.Ser384Gly/c.1150A>G (gaC/gaT) HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic NOTES ENSTO R PA	3 ACMG 0000037012 S 12417413 os 1:321313 ACMG 0000037012 S 12417413 os 1:321313	Fathnm Fathnm Scores Mutationtaster Mutationassessor Placentalphylop46 Polyphen2 Cadd Lof Nmd Fathnm Scores Mutationtaster Mutationtaster Mutationassessor Placentalphylop46 Polyphen2 Cadd Lof Primatephylop46 Polyphen2 Cadd Lof Nmd	0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32) 3.9 (11) -0.06 (5) - - 0.0 (46) 1.4 (30) 0.66 (21) 2.8 (15) 1.0 (32) 3.9 (11) -0.06 (5) -	Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001 Frequency 1000GPI Avr 0,05 Afr 0,01 Eur 0,11 Amr 0,09 Asn 0,0001	ESP6500 Ea 0.05 Aa 0.01 ESP6500 Ea 0.05 Aa 0.01	ExAc Avr 0,04 ExAc Avr 0,04

Printed report. Design passed user- and scale-tests with +10 databases.

Instrument: Illumina HiSeq Library: mate-pair Specimen: whole blood

Database version ClinVar 1.2, apr'15 CADD 1.2, jan'15 1000Genomes 0.3, jan'13

PolyPhen-2 2.2, feb'12 ExAC 0.3, dec'14



Variants report

Robert Hairullin

mutationtaster 0.99989 Af 0.00017 Aa - mutationassessor 2.275 Afr - Ea -	Af – Af –
ENST00000368089 Pos 1:160012175 mutationassessor rank 0.74639 Amr – Alspac fathmm -3.88 Adj 0.00017 Af – fathmm rank 0.95957 Eas – – cadd phred 23.5 Fin 0.0003 Twinsuk cadd phred 23.5 Fin 0.0003 Twinsuk cadd rank 0.72574 Nfe 0.00029 Af 0.0002' sift 0.059 Oth –	Amr – Eas – Eur – Sas –
phylop7way vertebrate 0.917 phylop7way vertebrate rank 0.60462	
Scores Frequency polyphen2 hvar 0.074 ExAc ESP6500 mutationtaster 0.99954 Af 0.027 Aa 0.00724	1000GP1 Af 0.012
ENST00000361951 mutationassessor 1.535 Afr 0.00654 Ea 0.03756 ENST00000361951 nutationassessor rank 0.49068 Amr 0.01641 Alspac fathmm -1.18 Adj 0.0266 Af 0.0319 fathmm rank 0.78323 Eas - - -	Afr 0.001 Amr 0.0230 Eas – Eur 0.0311
cadd phred 22.7 Fin 0.03009 Twinsuk cadd rank 0.67021 Nfe 0.03733 Af 0.03423 sift 0.078 Oth 0.0293 Af 0.03423 sift 0.33878 Sas 0.0153	Sas 0.012:
phastcons7way vertebrate 0.998 phastcons7way vertebrate rank0.72479 phylop7way vertebrate 0.917 phylop7way vertebrate rank 0.60462	
Scores Frequency	
mutationassessor 0.895 Afr 0.03834 Ea 0.0158	Af 0.024 Afr 0.048
ENST00000358025 Pos 14:64580075mutationassessor rank0.30057 (athmmAmr 0.00752 (athmmAlspac (Af 0.01453)fathmm0.38Adj0.01711 (athmm rank)Adj0.01711 (Af 0.01453)fathmmcadd phred2.59Fin0.02454Twinsuk (add rank)cadd rank0.07097Nfe0.01551Af0.0151 (Af 0.0151)sift0.093Oth0.02533Sift rank0.3149Sas0.02217phastcons7way vertebrate0.032 (phastcons7way vertebrate rank).124480.034Sas0.02217	Amr 0.017 Eas – Eur 0.0119 Sas 0.034
phylop7way vertebrate 0.991 phylop7way vertebrate rank 0.76621	
phylop7way vertebrate 0.991 phylop7way vertebrate rank 0.76621	

Read about exome analysis limitations on the last page

More variants on the p. 2 \rightarrow







About

Q Search by patient or report

New patient 0 panels, 12 exomes, 0 genomes

> Drag file here to upload or select from the list below

7bbaca6f-0e9b-11e5-918c-22000...

7bb6f687-0e9b-11e5-918c-2200...

sample_group (2 files)

test (2).fastq

- 454Reads.MID19.fq
- test (1) (2).fastq
- test (I) (I).fastq
- New folder (6 files)

test (1).fastq

Upload file 谷

50 Gb of 250 Gb

Patients

Select patient to view

7 may 2015 **Bob Stempsor**

3 may 2015 Emil Hakov

I may 2015 **Robert Hairul**

10 april 2015 Artur Karaiva

5 april 2015 Nick Stephan

25 march 2015 Artemy Adam

16 march 2015 Arnold Bishop

7 march2015

Patients. Uploading and results pages were united in a friendly one.

L001_1004_KJSC120914KJ216D.bam

and edit report	
in	
nskiy	
son	

ignat 1990@gmail.com







Filter

Depth and quality

Diseases and genes

Chromosome, position, rsID

Variant type, zygosity, effect

Scores

Populations frequency

Clinvar



253 variants, Robert Hairullin

UTS2 (SNP) Heterozygote non synonymous coding

p.Thr21Met/c.62C>T (aCg/aTg)

VWA5BI (SNP) Heterozygote non synonymous coding

p.Arg880His/c.2639G>A (cGc/cAc)

NBPF3 (SNP) Heterozygote non synonymous coding+splice site region

Web report. Layout and architecture followed the printed report logic.

 \bigcirc Search for disease in OMIM





About i

iBinom v2.1

Database version

Privacy agreement

Service agreement

Tell us about a problem via email

iBinom v2.1

Database version ClinVar 1.2, apr'15 CADD 1.0, may'14 HGMD 0.31, sept'14

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I. PERSONAL INFORMATION COLLECTED AND COLLECTION METHODS.

Items Subject to Collection:

Genomic Information. iBinom may collect and store the sequence data, reads type, file types and sizes, biases, errors, and trends within and across Data, instrument type and identifiers, analysis

Bonus. Even the service pages got a stylish look.

Powerful solution for genomic data analysis

PolyPhen 1.2, dec'14

ExAc 1.3, aug'13



Invention We invented the industry's new mutation frequency chart, allowing users to spot pathogenic mutations with a quick scan.



Pathogenic

Better to check

Mutation frequency is a set of small numbers 0.0001, 0.001, 0.1, 0.6. Plotting them on a linear scale will put all on zero. Adjusting the scale will make charts on different rows incomparable. Using arcsin we got to show difference while keeping the same scale.

Clearly pathogenic



Instrument: Illumina HiSeq Library: mate-pair Specimen: whole blood Bad scanned genes!

Database version ClinVar 1.2, apr'15,

Robert Hairullin, 27 august 1994, male Filter: DP>20, QUAL>20, Missense, Population frequency ExAc < 1%.

ADF. Homozygote non synonymous coding (SNP) p.Ser384Gly/c.1150A>G (gaC/gaT)

HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic

Wificat

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ADF. Homozygote non synonymous coding (SNP) p.Ser384Gly/c.1150A>G (gaC/gaT)

HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic

NOTES 50150

ENST

ADF. Homozygote non synonymous coding (SNP) p.Ser384Gly/c.1150A>G (gaC/gaT)

HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic

NOTES

ENST

(ADE) Homozygote non synonymous coding (SNP) p.Ser384Gly/c.1150A>G (gaC/gaT)

HGMD Adenosine, monophosp, hate, deaminase, deficency. Clinvar Patogenic*

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ENST

ADF. Homozygote non synonymous coding (SNP) Ser384Gly/c.1150A>G (gaC/gaT)

MD Adenosine, monophosp, hate, deaminase, deficency. rar Patogenic

ENST

about exome analysis limitations on the last page

PolyPhen 1.2, dec'14 CADD 1.0, may'14 ExAc 1.3, aug'13 HGMD 0.31, sept'14 1000Genomes 0.3, jan'13



BINOM

Ne13246578

1	Scores		Free	quency			and the second s	
4	Mutationtaster	0,0 (46)	100	OGPI	ES	P6500	ExAc	
ACMG	Mutationassessor	1,4 (30)	Avr	0.05	Ea	0,05	Avr 0,04	
	Placentalphylop46	0,66 (21)	Afr	0.01	Aa	0,01		
	Primatephylop46	2,8 (15)	Eur	0,11				
0000017013	Polyphen2	1,0 (32)	Amr	0.09				
1512417413	Cadd	(3,9 (11))	Asn	0,0001				
los 1 321313	Lof	-0.06 (5)						
	Nmd	-						
	Fathom	-						
1	Scores in man			Frequency				
4	Mutationtaster	0.0 (46)	1000	OGPI	ESF	6500	ExAc	
ACMG	Mutationassessor	1,4 (30)	Avr	0.05	Ea	0.05	Avr 0.04	
	Placentalphylop46	0.66 (21)	Afr	0,01	Aa	0.01		
	Primatephylop46	2,8 (15)	Eur	0.11				
	Polyphen2	1,0 (32)	Amr	0,09				
\$12417413	Cadd	3,9 (11)	Asn	0,0001				
os 1:321313	Lof	-0.06 (5)						
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	Primatechulosde	28(15)	Eur	0,01	A	0,01		
	Polyphen7	10(22)	Ame	0.00				
000037012	Cadd	3.9 (11)	Are	0,09				
5 12417413	Lof	3,7 (11)	Asn	0,0001				
a national	Nmd	-0,00 (3)						
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5	Mutationtaster	0,0 (46)	1000	GPI	ESP	6500	ExAc	
ACMG	Mutationassessor	1,4 (30)	Avr	0.05	Ea	0.05	Avr 0,04	
242.34	Placentalphylop46	0.66 (21)	Afr	0,01	Aa	0,01		
	Primatephylop46	2,8 (15)	Eur	0,11				
000037012	Polyphen2	1.0 (32)	Amr	0,09				
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ACMG	Mutationassessor	1,4 (30)	Avr	0.05	Ea	0,05	Avr 0.04	
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	Nmd	-					C. MAC	
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More mutation



Results Sales were unblocked–doctors were mentioning iBinom as the first usercentred DNA analysis tool.

User and scalability test passed. Team was ready to kick-start the sales on the upcoming conference. Unfortunately, startup was closed.

Special thanks to Andrey Afanasyev, Alexandra Vachnadze, Vasily Sitnik, Vladimir Naumov, Ilya Kitayev, and Aleksander Bespoyasov.

lestimonial



Andrei Afanasyev **CEO** and Founder at iBinom

Platform became solid, clear for doctors, and easy to use. Understanding bioinformatics in a couple weeks while managing the redesign is something l've never seen before



Nutipy Outputs with Bold design moves from your new Head of Designance mentor For startups, agencies, and designers. Shared at \$99.

With love.

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