Simplifying the DNA analysis SaaS in 4 weeks.

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

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.

Challenge

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	Alt	Zygosity	Gene	rsID	Transcript ID	Amino acid c	Quality	Depth	Effect	Variant type	Quality filter	Codon chang	lof	nmd	polyphen2_h	m	utationtast	mutationass	fathmm	sift	cadd	phylop46way	phylop46way	1000gp1_af	1000gp1_afr	1000gp1_eu	1000gp1_am	1000gp1_asr	esp6500_aa	esp6500_ea	HGMD	HGMD Phen	clinvar_signif
	100672060	T	C	hom	DBT	rs12021720	ENST000003	p.Ser384Gly/	230	391	NON_SYNON	SNP	PASS	Agt/Ggt			0		1		1.2	T	-0.69	-0.21	1.5	0.9	0.78	0.91	0.94	0.97	0.77	0.9	no		Pathogenic
	115231254	G	A	het	AMPD1	rs61752479	ENST000005	p.Pro81Leu/c	220	447	NON_SYNON	SNP	PASS	cCg/cTg			1		0	1.4	-0.06	D	3.9	0.66	2.8	0.054	0.0061	0.11	0.086	0	0.023	0.13	yes	Adenosine m	Pathogenic
	115236057	G	A	het	AMPD1	rs17602729	ENST000005	p.Gln45*/c.1	220	207	STOP_GAINE	SNP	PASS	Caa/Taa	1	1			0				5	0.65	1.1	0.054	0.0061	0.11	0.083	0	0.024	0.13	yes	Adenosine m	Pathogenic
	171080080	G	A	het	FMO3	rs1736557	ENST000003	p.Val257Met	210	40	NON_SYNON	SNP	PASS	Gtg/Atg			0.0073		1	0.36	0.58	T	1.2	-1.2	-2.2	0.094	0.028	0.058	0.094	0.2	0.037	0.069	yes	FMO3 varian	Pathogenic
	196659237	C	T	het	CFH	rs1061170	ENST000003	p.His402Tyr/	220	242	NON_SYNON	SNP	PASS	Cat/Tat			0.0005		1		-0.03	T	-2.3	-4.4	-4	0.72	0.62	0.63	0.73	0.93	0.64	0.62	no		Pathogenic
	203194186	C	T	het	CHIT1	rs2297950	ENST000003	p.Gly102Ser/	220	79	NON_SYNON	SNP	PASS	Ggc/Agc			1		0	1.7	3.8	D	4	0.66	0.32	0.3	0.29	0.32	0.29	0.3	0.27	0.29	yes	Colorectal ca	Pathogenic
	231408091	A	G	het	GNPAT	rs11558492	ENST000003	p.Asp519Gly	220	35	NON_SYNON	SNP	PASS	gAt/gGt			0.031		1	1	-0.11	D	1.8	0.46	-0.13	0.14	0.075	0.19	0.2	0.1	0.092	0.21	no		Pathogenic
1	51549496	T	C	het	MSMB	rs10993994	ENST000003	c.-88T>C	220	41	UPSTREAM	SNP	PASS																				no		Pathogenic
1	70645376	A	C	het	STOX1	rs10509305	ENST000002	p.Glu608Asp	220	35	NON_SYNON	SNP	PASS	gaA/gaC			0		1	-1.9	-0.81	T	-2.8	-2.5	-0.04	0.14	0.028	0.21	0.24	0.096	0.058	0.21	yes	Preeclampsia	Pathogenic
1	18290859	C	T	hom	SAA1	rs1136743	ENST000003	p.Ala70Val/c	190	31	NON_SYNON	SNP	PASS	gCc/gTc			0.006		1			3	T	-0.34	-0.39	0.003	0.48	0.45	0.61	0.58	0.26			no		Pathogenic
1	122295335	T	C	hom	HPD	rs1154510	ENST000002	p.Thr33Ala/c	230	55	NON_SYNON	SNP	PASS	Acg/Gcg					0		-0.38	T	0.93	-0.23	1.5	0.87	0.95	0.88	0.8	0.83	0.96	0.87	no		Pathogenic
1	94847415	A	G	het	SERPINA1	rs6647	ENST000003	p.Val237Ala/	100	39	NON_SYNON	SNP	PASS	gTg/gCg			0		1	-0.36	-1.8	T	0.65	-2.8	-2.2	0.24	0.63	0.22	0.13	0.024	0.54	0.21	no		Pathogenic
1	28260053	G	A	het	OCA2	rs1800401	ENST000003	p.Arg305Trp/	220	41	NON_SYNON	SNP	PASS	Cgg/Tgg			0.42	0	08		1.6	-2.7	D	1.7	-0.77	0.73	0.063	0.13	0.061	0.072	0	0.13	0.05	yes	Darker eye c	Pathogenic
1	27374180	T	C	het	IL4R	rs1805015	ENST000001	p.Ser503Pro/	220	34	NON_SYNON	SNP	PASS	Tcc/Ccc			0.12		1	1.1		3	T	2.1	0.46	0.76	0.21	0.46	0.17	0.17	0.075	0.36	0.16	yes	Atopy, assoc	Pathogenic
1	56548501	C	T	hom	BBS2	rs4784677	ENST000002	p.Ser70Asn/c	230	322	NON_SYNON	SNP	PASS	aGc/aAc					1		-1.2	T	0.72	-0.33	0.95	1	1	0.99	1	1	1	0.99	no		Pathogenic
1	3397702	C	T	het	ASPA	rs12948217	ENST000002	p.Tyr231Tyr/	220	314	SYNONYMOU	SNP	PASS	taC/taT																			no		Pathogenic
1	7125591	T	C	het	ACADVL	rs113994167	ENST000005	p.Val306Ala/	220	377	NON_SYNON	SNP	PASS	gTg/gCg			0.39		1	0.95	-5.3	D	4.2	0.53	2						0.00023	0.0013	no		Pathogenic
1	42453065	A	C	hom	ITGA2B	rs5911	ENST000002	p.Ile874Ser/c	230	22	NON_SYNON	SNP	PASS	aTc/aGc			0		0	-0.69	1.1	T	1.7	-0.32	1	0.4	0.43	0.4	0.36	0.42	0.39	0.37	yes	Reduced pos	Pathogenic
1	45360730	T	C	het	ITGB3	rs5918	ENST000005	p.Leu59Pro/c	220	434	NON_SYNON	SNP	PASS	cTg/cCg			0.005		1	1.4		3	T	0.49	-1.1	-4	0.092	0.12	0.13	0.097	0.0087	0.11	0.15	yes	Neonatal allo	Pathogenic
2	46931109	G	A	het	COL18A1	rs12483377	ENST000003	p.Asp1675As	220	515	NON_SYNON	SNP	PASS	Gac/Aac			1		0	1.6	0.6	T	4.2	0.66	2.5	0.055	0.02	0.086	0.072	0.033	0.036	0.088	no		Pathogenic
2	18901004	C	T	hom	PRODH	rs450046	ENST000003	p.Arg521Gln	230	33	NON_SYNON	SNP	PASS	cGg/cAg			0		0		1.6	T	1.6	-0.47	0.75	0.91	0.8	0.93	0.95	0.96	0.84	0.94	no		Pathogenic
	182759467	T	G	het	MCCC1	rs119103213	ENST000002	p.Arg385Ser/	220	468	NON_SYNON	SNP	PASS	agA/agC			0.88		1	4.8		-4	D	3.3	-0.39	-0.14								yes	3-methylcrot	Pathogenic

Position	Ref	Alt	Zygosity	Gene	rsID	Transcript ID	Amino acid c	Quality	Depth	Effect	Variant type	Quality filter	Codon chang	lof	nmd	polyphen2_h	m
100672060	T	C	hom	DBT	rs12021720	ENST000003	p.Ser384Gly/	230	391	NON_SYNON	SNP	PASS	Agt/Ggt			0	
115231254	G	A	het	AMPD1	rs61752479	ENST000005	p.Pro81Leu/c	220	447	NON_SYNON	SNP	PASS	cCg/cTg			1	
115236057	G	A	het	AMPD1	rs17602729	ENST000005	p.Gln45*/c.1	220	207	STOP_GAINE	SNP	PASS	Caa/Taa	1	1		
171080080	G	A	het	FMO3	rs1736557	ENST000003	p.Val257Met	210	40	NON_SYNON	SNP	PASS	Gtg/Atg			0.0073	
196659237	C	T	het	CFH	rs1061170	ENST000003	p.His402Tyr/	220	242	NON_SYNON	SNP	PASS	Cat/Tat			0.0005	
203194186	C	T	het	CHIT1	rs2297950	ENST000003	p.Gly102Ser/	220	79	NON_SYNON	SNP	PASS	Ggc/Agc			1	
231408091	A	G	het	GNPAT	rs11558492	ENST000003	p.Asp519Gly	220	35	NON_SYNON	SNP	PASS	gAt/gGt			0.031	
51549496	T	C	het	MSMB	rs10993994	ENST000003	c.-88T>C	220	41	UPSTREAM	SNP	PASS					
70645376	A	C	het	STOX1	rs10509305	ENST000002	p.Glu608Asp	220	35	NON_SYNON	SNP	PASS	gaA/gaC			0	
18290859	C	T	hom	SAA1	rs1136743	ENST000003	p.Ala70Val/c	190	31	NON_SYNON	SNP	PASS	gCc/gTc			0.006	
122295335	T	C	hom	HPD	rs1154510	ENST000002	p.Thr33Ala/c	230	55	NON_SYNON	SNP	PASS	Acg/Gcg				
94847415	A	G	het	SERPINA1	rs6647	ENST000003	p.Val237Ala/	100	39	NON_SYNON	SNP	PASS	gTg/gCg			0	
28260053	G	A	het	OCA2	rs1800401	ENST000003	p.Arg305Trp/	220	41	NON_SYNON	SNP	PASS	Cgg/Tgg			0.42	0.
27374180	T	C	het	IL4R	rs1805015	ENST000001	p.Ser503Pro/	220	34	NON_SYNON	SNP	PASS	Tcc/Ccc			0.12	
56548501	C	T	hom	BBS2	rs4784677	ENST000002	p.Ser70Asn/c	230	322	NON_SYNON	SNP	PASS	aGc/aAc				
3397702	C	T	het	ASPA	rs12948217	ENST000002	p.Tyr231Tyr/	220	314	SYNONYMOU	SNP	PASS	taC/taT				
7125591	T	C	het	ACADVL	rs113994167	ENST000005	p.Val306Ala/	220	377	NON_SYNON	SNP	PASS	gTg/gCg			0.39	
42453065	A	C	hom	ITGA2B	rs5911	ENST000002	p.Ile874Ser/c	230	22	NON_SYNON	SNP	PASS	aTc/aGc			0	
45360730	T	C	het	ITGB3	rs5918	ENST000005	p.Leu59Pro/c	220	434	NON_SYNON	SNP	PASS	cTg/cCg			0.005	
46931109	G	A	het	COL18A1	rs12483377	ENST000003	p.Asp1675As	220	515	NON_SYNON	SNP	PASS	Gac/Aac			1	
18901004	C	T	hom	PRODH	rs450046	ENST000003	p.Arg521Gln	230	33	NON_SYNON	SNP	PASS	cGg/cAg			0	
182759467	T	G	het	MCCC1	rs119103213	ENST000002	p.Arg385Ser/	220	468	NON_SYNON	SNP	PASS	agA/agC			0.88	

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

BINOM Beta

About Service agreement Manual info@ibinom.com

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

Results **Raw data** (43.22 Gb of 250.00 Gb)

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

Group Move to archive Delete

Archive (0 files) 0 bytes

018_1.fastq.gz 1.41 Gb

018_2.fastq.gz 1.40 Gb

454Reads.MID19.fq 3.63 Mb

Sort by name

Press to upload or drag'n'drop

Analyze

Homescreen & upload. Unclear hierarchy, navigation and layout.

BINOM Beta

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ignat1990@gmail.com (Sign out)

0 panels, 16 exomes, 0 genomes

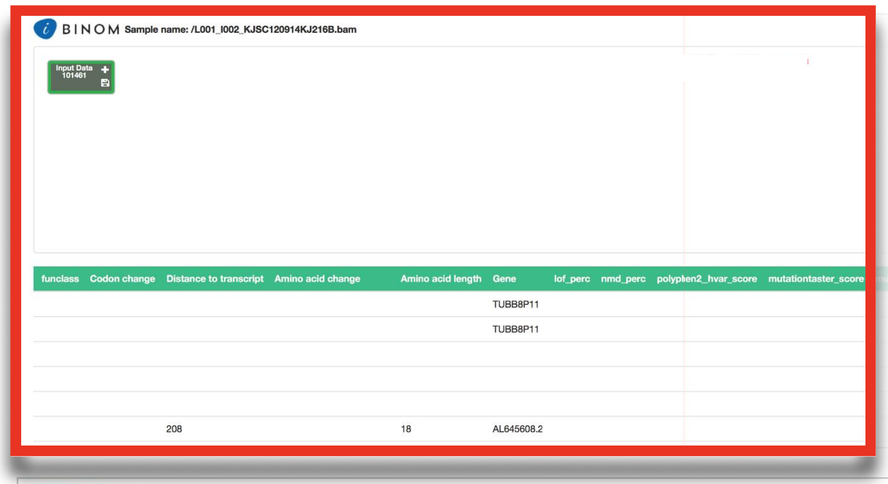
Raw data **Results**

You can download result files here

Move to archive | Delete

Sample name	Aligned reads (bam + bai)	VCF Filter	Report	Variant call (vcf)	Variant call (csv)	Date
Archive (16 files)						
L001_I002_KJSC120914KJ216B.bam	Filter	3.48 Mb	25.51 Mb	14.40 Mb		07.04.2015 03:49:53
sample.fastq.gz (7)	Filter	745.47 Kb	19.85 Mb	8.82 Mb		07.05.2015 02:01:20
sample.fastq.gz (8)	Filter	745.46 Kb	19.85 Mb	8.82 Mb		08.05.2015 20:40:08
sample.fastq.gz (9)		718.35 Kb	0 bytes	0 bytes		13.05.2015 13:56:09
sample.fastq.gz (10)	Filter	744.97 Kb	20.01 Mb	8.82 Mb		13.05.2015 20:54:12
Myodystrophy_M	Filter	745.35 Kb	42.46 Mb	19.51 Mb		29.05.2015 19:50:09
short.fastq.gz (1)	Filter	746.90 Kb	33.42 Kb	12.95 Kb		02.06.2015 16:00:26
001.fastq	Filter	744.88 Kb	90.87 Kb	41.42 Kb		04.06.2015 13:43:53
sample.fastq.gz (6)	Filter	745.46 Kb	19.85 Mb	8.82 Mb		06.05.2015 14:34:09
Pomykalov	Filter	745.41 Kb	17.36 Mb	8.29 Mb		09.06.2015 18:38:51
Zernov 018 deafness	Filter	745.14 Kb	33.16 Mb	15.49 Mb		09.06.2015 23:06:10
Zernov 002 hypotrihosis	Filter	745.10 Kb	31.27 Mb	14.57 Mb		14.06.2015 01:48:50

Processed results. Unclear, inconsistent and complex structure.



BINOM Sample name: /L001_l002_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	mutationassessor_score
					TUBB8P11					
					TUBB8P11					
		208		18	AL645608.2					

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

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
Iterating on the optimal data-visualisation and look.

Web design
Applying new ideas from the reports. Drafting the architecture.

Frontend-development
Delivering the ready parts and reviewing the prototypes.

Mutations

ADF: Non synonymous coding
Chr-1, position-123123, hom (ENST 0000037012)

p.Ser384Gly/c.1150A>G
G→A RS12417413

Clinvar: patogenic
HKMD: adenosine, monophos,hate, deaminase, deficiency

ACMG-7

Lof	Nmd	Poly phen2 gvar	Fath mm	Sift	Cadd	Phylop 46way primate	Phylop 46way placental	Mutation taster	Mutation assessor	1000gp1 average	1000gp1 afr	1000gp1 eur	1000gp1 amr	1000gp1 asn	esp600 aaaf	esp600 eaaf
1	0,1	0,5	0,8	1	0,1	0,3	0,5	0,8	0,2	0,03	0,05	0,03	0,02	0,02	0,02	0,02

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1	0,1	0,5	0,8	1	0,1	0,3	0,5	0,8	0,2	0,03	0,05	0,03	0,02	0,02	0,02	0,02

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1	0,1	0,5	0,8	1	0,1	0,3	0,5	0,8	0,2	0,03	0,05	0,03	0,02	0,02	0,02	0,02

WASH7P: EXON
n.1493G>A
no:
Chr-1, position-14653, het
ENST00000438504, RS375086259

Scores Clinvar-, ACMG-7
fathmm | mutation taster | primate phylop46 | poly phen2 | mutation assessor | placental phylop46 | cadd

Frequency 0,01–0,0001
1000GP1 avr afr eur amr asn ESP6500 ea aa

DDX11L1: DOWNSTREAM
n.*1657A>G
no:
Chr-1, position-14907, hom
ENST00000456328, RS79585140

Scores Clinvar-, ACMG-7
fathmm | mutation taster | primate phylop46 | poly phen2 | mutation assessor | placental phylop46 | cadd

Frequency 0,01–0,0001
1000GP1 avr afr eur amr asn ESP6500 ea aa

DDX11L1: DOWNSTREAM
n.*1657A>G
no:
Chr-1, position-14930, hom
ENST00000456328, RS75454623

Scores Clinvar-, ACMG-7
fathmm | mutation taster | primate phylop46 | poly phen2 | mutation assessor | placental phylop46 | cadd

Frequency 0,01–0,0001
1000GP1 avr afr eur amr asn ESP6500 ea aa

DDX11L1: DOWNSTREAM
n.*1657A>G

Scores Clinvar-, ACMG-7
fathmm | mutation taster | primate phylop46 | poly phen2 | mutation assessor | placental phylop46 | cadd

Mutations

Gene and effect
ADF homozygote non synonymous coding
Chr-1, position-123123 (ENST 0000037012)

Status
Clinvar: patogenic
HKMD: adenosine, monophos, hate, deaminase, deficiency

Acid change
p.Ser384Gly/c.1150A>G
G→A RS12417413

ACMG
7

Lof	Nmd	Polyphen2 gvar	Fathmm	Sift	Cadd	Phylop 46way primate	Phylop 46way pacentai	Mutation taster	Mutation asessor	1000gp1 average	1000gp1 afr	1000gp1 eur	1000gp1 amr	1000gp1 asn	esp600 aaaf	esp600 eaaf
1	0,1	0,5	0,8	1	0,1	0,3	0,5	0,8	0,2	0,03	0,05	0,03	0,02	0,02	0,02	0,02

Frequency (0,1–0,0001)
1000GP1 avr 0,05 afr 0,01 eur 0,11 amr 0,09 asn 0,00
ESP6500 aa 0,09 ea 0,00

Scores (ACMG-7)
-0,06 fathnm | 0,0 mutation taster | 0,66 primate phylop46 | 1,0 poly phen2 | 1,4 mutation asessor | 2,8 placental phylop46 | 3,9 cadd

ADF: Non synonymous coding
Chr-1, position-123123, homozygote

Patogenic
Clinvar

Adenosine, monophos, hate, deaminase, deficiency
HKMD

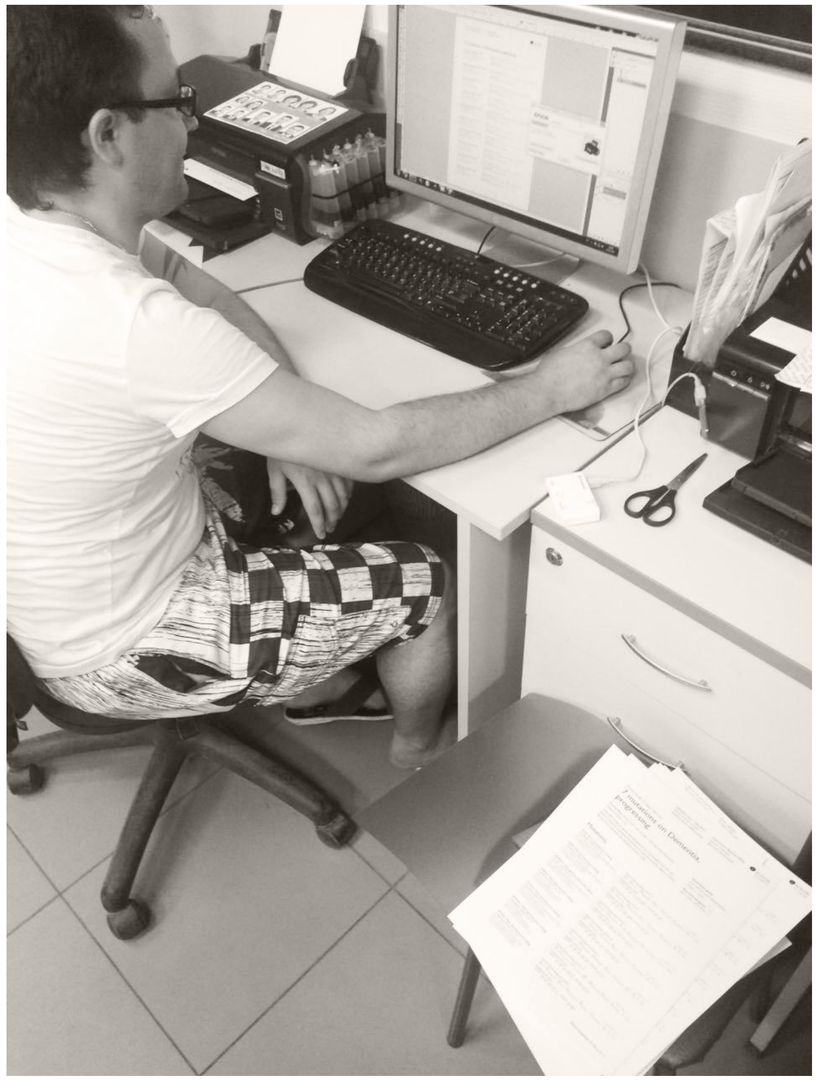
ENST0000037012, RS12417413

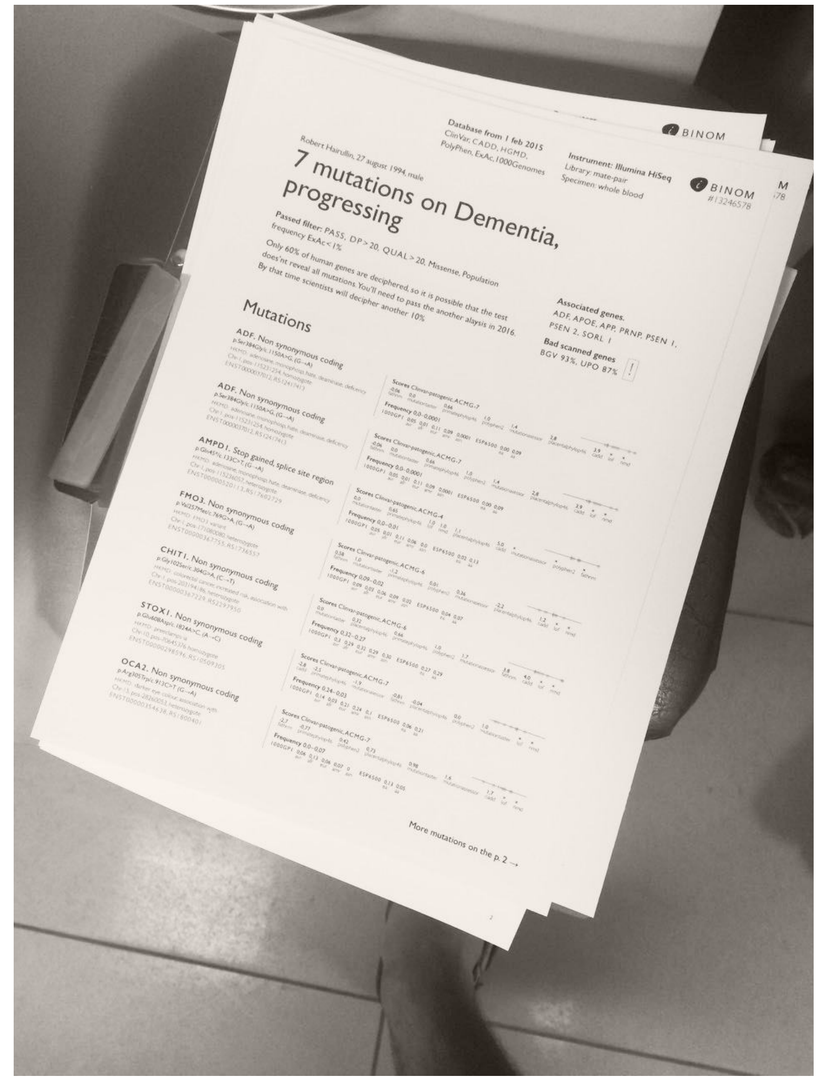
Frequency (0,00–0,0001)
1000GP1 0,05 avr | 0,01 afr | 0,11 eur | 0,09 amr | 0,0001 asn | ESP6500 0,00 ea | 0,09 aa

Scores (ACMG-7)
-0,06 fathnm | 0,0 mutation taster | 0,66 primate phylop46 | 1,0 poly phen2 | 1,4 mutation asessor | 2,8 placental phylop46 | 3,9 cadd

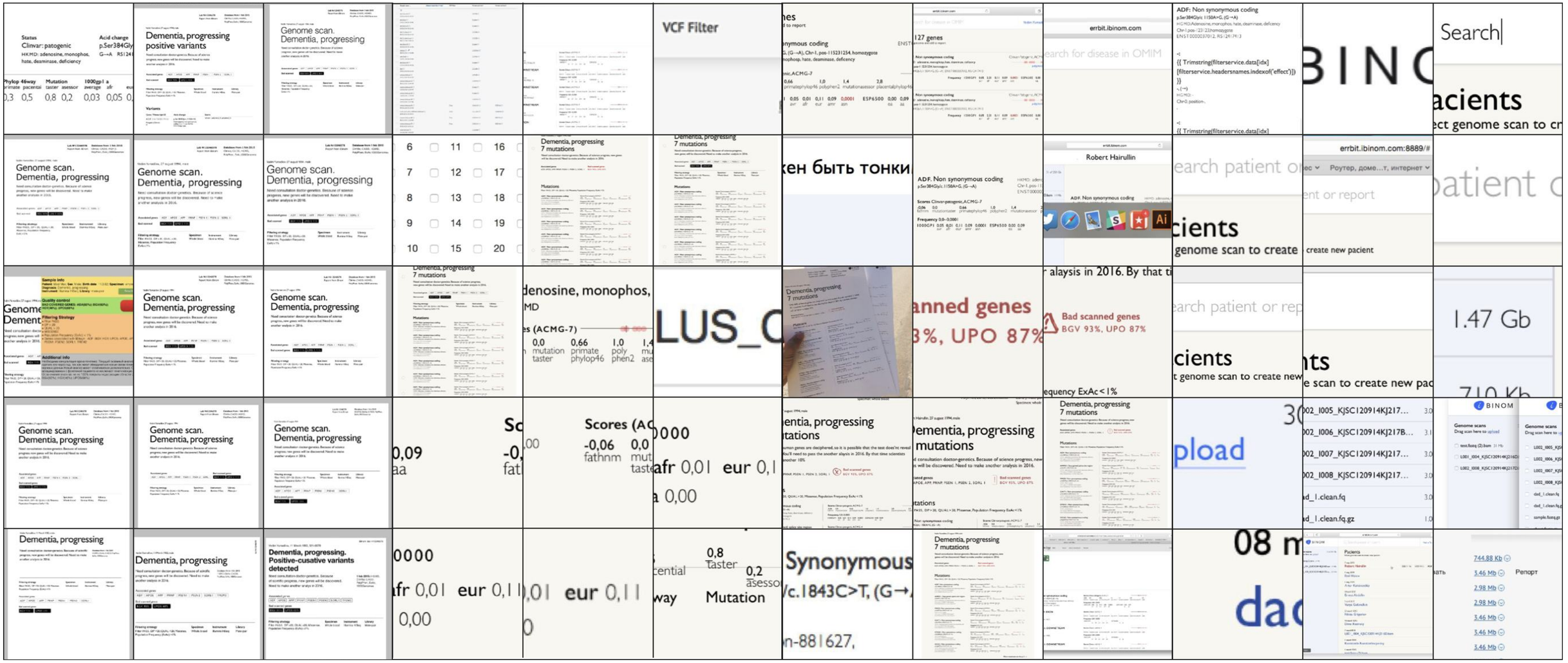
Amino-acid change
p.Ser384Gly/c.1150A>G
(G→A)

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





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



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

Ready.

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, 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
#13246578

Dementia, progressing. Mutations report

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

ADF. Homozygote non synonymous coding (SNP) — 4 ACMG
p.Ser384Gly/c.1150A>G (gaC/gaT)
HGMD Adenosine, monophosp, hate, deaminase, deficency.
Clinvar Patogenic
NOTES
ENST00000037012
RS12417413
Pos 1:321313

Scores		Frequency			
Mutationtaster	0,0 (46)	1000GPI		ESP6500	ExAc
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		
Cadd	3,9 (11)	Asn	0,0001		
Lof	-0,06 (5)				
Nmd	–				
Fathnm	–				

ADF. Homozygote non synonymous coding (SNP) — 4 ACMG
p.Ser384Gly/c.1150A>G (gaC/gaT)
HGMD Adenosine, monophosp, hate, deaminase, deficency.
Clinvar Patogenic
NOTES
ENST00000037012
RS12417413
Pos 1:321313

Scores		Frequency			
Mutationtaster	0,0 (46)	1000GPI		ESP6500	ExAc
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		
Cadd	3,9 (11)	Asn	0,0001		
Lof	-0,06 (5)				
Nmd	–				
Fathnm	–				

ADF. Homozygote non synonymous coding (SNP) — 5 ACMG
p.Ser384Gly/c.1150A>G (gaC/gaT)
HGMD Adenosine, monophosp, hate, deaminase, deficency.
Clinvar Patogenic
NOTES
ENST00000037012
RS12417413
Pos 1:321313

Scores		Frequency			
Mutationtaster	0,0 (46)	1000GPI		ESP6500	ExAc
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Polyphen2	1,0 (32)	Amr	0,09		
Cadd	3,9 (11)	Asn	0,0001		
Lof	-0,06 (5)				
Nmd	–				
Fathnm	–				

ADF. Homozygote non synonymous coding (SNP) — 3 ACMG
p.Ser384Gly/c.1150A>G (gaC/gaT)
HGMD Adenosine, monophosp, hate, deaminase, deficency.
Clinvar Patogenic
NOTES
ENST00000037012
RS12417413
Pos 1:321313

Scores		Frequency			
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Mutationassessor	1,4 (30)	Avr	0,05	Ea 0,05	Avr 0,04
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Polyphen2	1,0 (32)	Amr	0,09		
Cadd	3,9 (11)	Asn	0,0001		
Lof	-0,06 (5)				
Nmd	–				
Fathnm	–				

ADF. Homozygote non synonymous coding (SNP) — 1 ACMG
p.Ser384Gly/c.1150A>G (gaC/gaT)
HGMD Adenosine, monophosp, hate, deaminase, deficency.
Clinvar Patogenic
NOTES
ENST00000037012
RS12417413
Pos 1:321313

Scores		Frequency			
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Polyphen2	1,0 (32)	Amr	0,09		
Cadd	3,9 (11)	Asn	0,0001		
Lof	-0,06 (5)				
Nmd	–				
Fathnm	–				

Bad scanned genes
CDH23

Selected genes
CDH23
CLDN14
COL11A2
DFNB31
DFNB59
ESPN
ESRRB
FOXI1
GIPC3
GJB2
GJB3
GJB6
GPSM2
GRXCR1
HGF
ILDR1
ADF
KCNJ10
LHFPL5
LOXHD1
LRTOMT
MARVELD2
MSRB3
MYO15A
MYO3A
MYO6
MYO7A
OTOA
OTOF
PCDH15
POU3F4
PRPS1
PTPRQ
RDX|
GJB3
GJB6
GPSM2
GRXCR1
HGF
ILDR1
KCNJ10
LHFPL5
LOXHD1
LRTOMT
MARVELD2
ILDR1
KCNJ10
LHFPL5
LOXHD1

Read about exome analysis limitations on the last page
More mutations on the p. 2 →

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

Database version
ClinVar 1.2, apr'15 PolyPhen-2 2.2, feb'12
CADD 1.2, jan'15 ExAC 0.3, dec'14
1000Genomes 0.3, jan'13

BINOM
№105

Variants report

Robert Hairullin

KCNJ10. Non synonymous coding
p.Leu50Phe/c.148C>T, (G→A)
Clinvar
NOTES
ENST00000368089
Pos 1:160012175

Scores		Frequency			
polyphen2 hvar	1	ExAc		ESP6500	1000GPI
mutationtaster	0.99989	Af	0.00017	Aa –	Af –
mutationassessor	2.275	Afr	–	Ea –	Afr –
mutationassessor rank	0.74639	Amr	–	Alspac	Amr –
fathmm	-3.88	Adj	0.00017	Af –	Eas –
fathmm rank	0.95957	Eas	–	Twinsuk	Eur –
cadd phred	23.5	Fin	0.0003	Af 0.00027	Sas –
cadd rank	0.72574	Nfe	0.00029		
sift	0.059	Oth	–		
sift rank	0.37489	Sas	–		
phastcons7way vertebrate	1				
phastcons7way vertebrate rank	0.90892				
phylop7way vertebrate	0.917				
phylop7way vertebrate rank	0.60462				

DARS2. Non synonymous coding
p.Gly338Glu/c.1013G>A, (G→A)
Clinvar
NOTES
ENST00000361951
Pos 1:173808677

Scores		Frequency			
polyphen2 hvar	0.074	ExAc		ESP6500	1000GPI
mutationtaster	0.99954	Af	0.027	Aa 0.00726	Af 0.01238
mutationassessor	1.535	Afr	0.00654	Ea 0.03756	Afr 0.00151
mutationassessor rank	0.49068	Amr	0.01641	Alspac	Amr 0.02305
fathmm	-1.18	Adj	0.0266	Af 0.03191	Eas –
fathmm rank	0.78323	Eas	–	Twinsuk	Eur 0.03181
cadd phred	22.7	Fin	0.03009	Af 0.03425	Sas 0.01227
cadd rank	0.67021	Nfe	0.03733		
sift	0.078	Oth	0.0291		
sift rank	0.33878	Sas	0.0153		
phastcons7way vertebrate	0.998				
phastcons7way vertebrate rank	0.72479				
phylop7way vertebrate	0.917				
phylop7way vertebrate rank	0.60462				

SYNE2. Non synonymous coding
p.Ile4209Thr/c.12626T>C, (T→C)
Clinvar; Likely benign
NOTES
ENST00000358025
Pos 14:64580075

Scores		Frequency			
polyphen2 hvar	0.005	ExAc		ESP6500	1000GPI
mutationtaster	1	Af	0.017	Aa 0.03473	Af 0.02436
mutationassessor	0.895	Afr	0.03834	Ea 0.01581	Afr 0.04841
mutationassessor rank	0.30057	Amr	0.00752	Alspac	Amr 0.01729
fathmm	0.38	Adj	0.01711	Af 0.01453	Eas –
fathmm rank	0.57592	Eas	0.00058	Twinsuk	Eur 0.01193
cadd phred	2.59	Fin	0.02454	Af 0.0151	Sas 0.03476
cadd rank	0.07097	Nfe	0.01551		
sift	0.093	Oth	0.02533		
sift rank	0.3149	Sas	0.02217		
phastcons7way vertebrate	0.032				
phastcons7way vertebrate rank	0.12448				
phylop7way vertebrate	0.991				
phylop7way vertebrate rank	0.76621				

Read about exome analysis limitations on the last page
More variants on the p. 2 →

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

BINOM About

Search by patient or report

ignat1990@gmail.com

New patient 50 Gb of 250 Gb

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 (1) (1).fastq
- New folder (6 files)
- test (1).fastq

Upload file

Patients

Select patient to view and edit report

7 may 2015
Bob Stempson

3 may 2015
Emil Hakov

1 may 2015
Robert Hairullin

10 april 2015
Artur Karaivanskiy

5 april 2015
Nick Stephan

25 march 2015
Artemy Adamson

16 march 2015
Arnold Bishop

7 march2015
L001_I004_KJSC120914KJ216D.bam

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

Filter

- Depth and quality
- Diseases and genes
- Chromosome, position, rsID
- Variant type, zygosity, effect
- Scores
- Populations frequency
- Clinvar

253 variants, Robert Hairullin

Create report

BENIGN PATHOGENIC

UTS2 (SNP)
Heterozygote non synonymous coding

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

Scores
MT psC7vr CDDp F.MMr PP3V pIP7vr

Frequency
k k_afr k_amr k_eas k_eur k_sas a e_ aa e_ea x x_afr x_amr x_adj x_eas x_

ENST00000054668, rs228648
pos: 1:7913430

VWA5B1 (SNP)
Heterozygote non synonymous coding

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

Scores
MT psC7vr CDDp F.MMr PP3V MAr SFTr

Frequency
k k_afr k_amr k_eas k_eur k_sas a e_ aa e_ea x x_afr x_amr x_adj x_eas x_

ENST00000375079, rs11582960
pos: 1:20671961

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

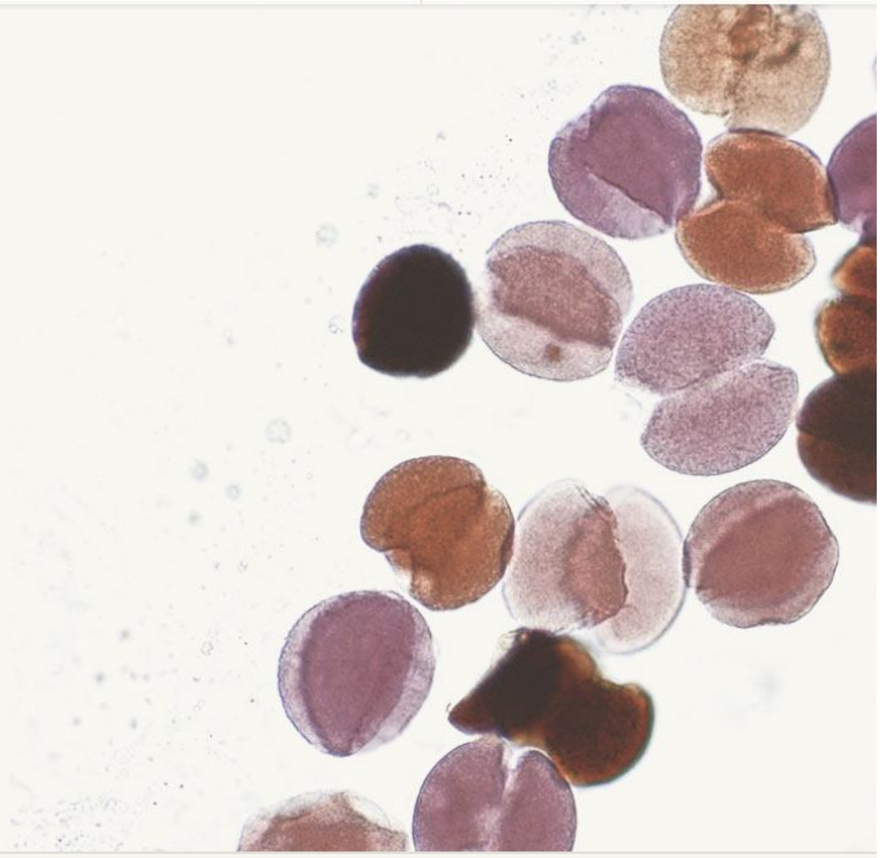
Scores
MT psC7vr CDDp F.MMr PP3V MAr SFTr

Frequency
k k_afr k_amr k_eas k_eur k_sas a e_ aa e_ea x x_afr x_amr x_adj x_eas x_

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

iBinom v2.1

Powerful solution for genomic data analysis



Database version

ClinVar 1.2, apr'15	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

Privacy agreement

This privacy policy applies to the services, products, technologies related to iBinom. It explains how and for what purposes iBinom may collect, use its customer's personal information and how to protect such information from you. Your use of the iBinom is Subject to this privacy policy, if you don't agree with any of the terms and conditions of this Privacy Policy, please do not submit your data or otherwise use the iBinom.

1. 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.

Invention

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

Pathogenic **Better to check** **Clearly pathogenic**

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.

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

Database version
ClinVar 1.2, apr'15, 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
№13246578

Dementia, progressing. Mutations report

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
4 ACMG

NOTES critical
ENST0000037012
RS12417413
Pos 1:321313

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Clinvar Patogenic
4 ACMG

NOTES 50150
ENST0000037012
RS12417413
Pos 1:321313

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Clinvar ~~Patogenic~~
3 ACMG

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Bad scanned genes
CDH23

Selected genes
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DFNB31
DFNB59
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ESRRB
FOXI1
GIPC3
GJB2
GJB3
GJB6
GPSM2
GRXCR1
HGF
ILDR1
ADF
KCNJ10
LHFPL5
LOXHD1
LRTOMT
MARVELD2
MSRB3
MYO15A
MYO3A
MYO6
MYO7A
OTOA
OTOF
PCDH
POU3
PRPS
PTP
RD
GJ
G
C

d about exome analysis limitations on the last page

More mutation

Results

Sales were unblocked—doctors were mentioning iBinom as the first user-centred 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.

Testimonial

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 I've never seen before

Multiply Outputs with Bold design moves from your new Head of Design and mentor.

For startups, agencies, and designers.

Shared at $99.

With love.

*

KK