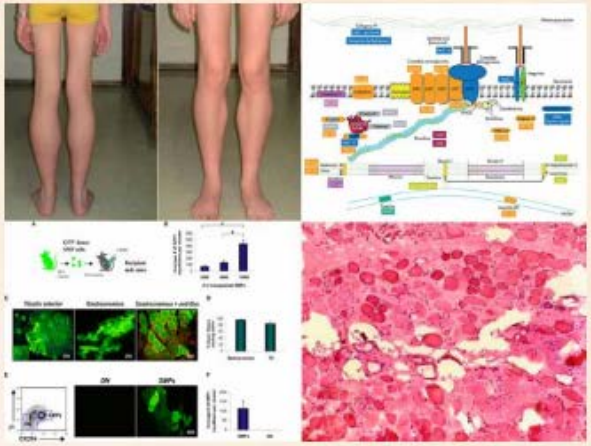
CURSO DE FORMACIÓN CONTINUADA
de las Profesiones Sanitarias de la Comunidad de Madrid – 1,9 Créditos

Hospital Universitario Ramón y Cajal
SaludMadrid
Comunidad de Madrid
Universidad de Alcalá

Enfermedades Musculares en la Infancia y Adolescencia (XIII)

Organizado por:
instituto ramón y cajal de investigación sanitaria irycis



Servicio de Pediatría. B. García Cuartero. Jefe de Servicio

17 y 18 de Marzo de 2016

Salón de Actos. Planta 0 centro.
Hospital Universitario Ramón y Cajal

Inscripción: GRATUITA, enviando un correo electrónico indicando nombre y filiación a glorenzosanz@yahoo.es

Coordinadores:

Dr. G. Lorenzo Sanz	Neurología Infantil S. de Pediatría.
Dra. R. Buenache Espartosa	Neurología Infantil S. de Pediatría.

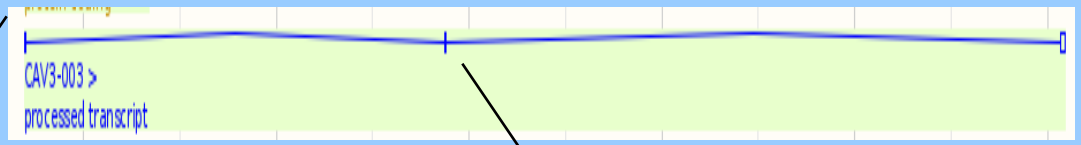
instituto ramón y cajal de investigación sanitaria irycis

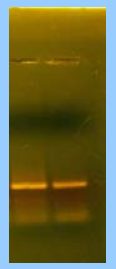
Curso de Formación Continuada de las Profesiones Sanitarias de la Comunidad de Madrid – 1,9 Créditos
(nº de expediente: *en trámite*)

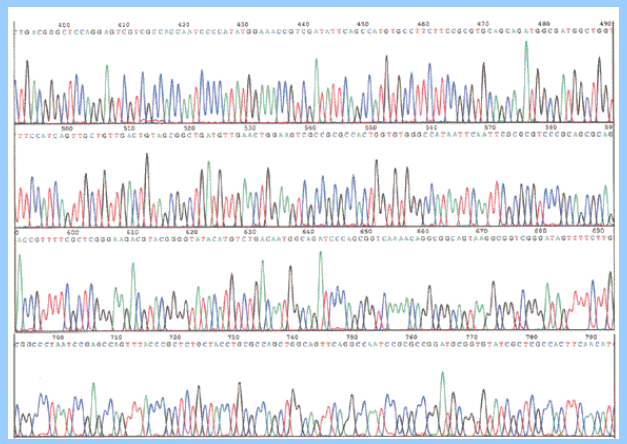
JUEVES 17 de Marzo

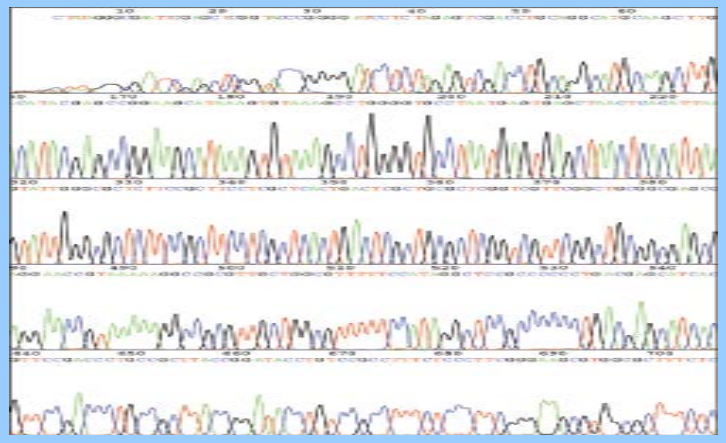
8:30	RECOGIDA DE DOCUMENTACIÓN
9:25	PRESENTACIÓN
9:30	**MESA REDONDA: Miopatías en la infancia y adolescencia (I)** **Moderador:** Dra. M.T. de Santos Moreno
	Evaluación del neonato y del lactante con hipotonía Dr. M. Madruga Garrido
	Valoración del paciente con sospecha de enfermedad neuromuscular. Algoritmos diagnósticos Dr. J. Esteban Pérez
10:30	COLOQUIO
10:45	**MESA REDONDA: Miopatías en la infancia y adolescencia (II)** **Moderador:** Dr. P. Castro de Castro
	Miopatías congénitas: correlación clínico–patológica genética Dr. A. Hernández Laín
	Contribución de la RM al diagnóstico de las enfermedades musculares Dra. F. Munell Casadesus
11:45	COLOQUIO
12:00	DESCANSO (café)
12:30	INAUGURACIÓN OFICIAL
	MESA REDONDA: Miopatías en la infancia y adolescencia (III) **Moderador:** Dra. C. Hernández Chico
	Estado actual de los ensayos clínicos en AME Dr. E. Tizzano Ferrari
	La secuenciación de nueva generación (NGS) en las enfermedades neuromusculares Dr. A. Jiménez Escrig
	Aportación de la inmunología al diagnóstico de las enfermedades neuromusculares Dra. A. Carrasco Sayalero
14:15	COLOQUIO
14:30	DESCANSO

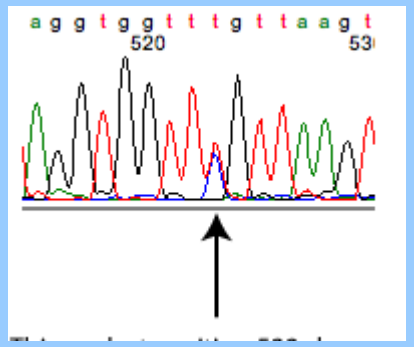
SECUENCIACION SANGER



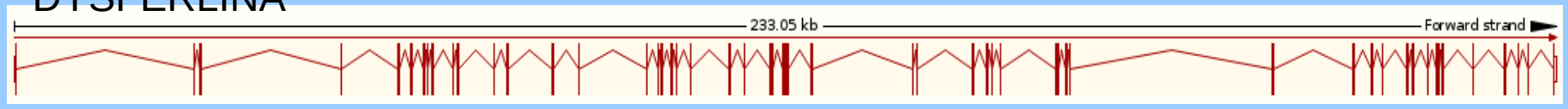








DYSFERLINA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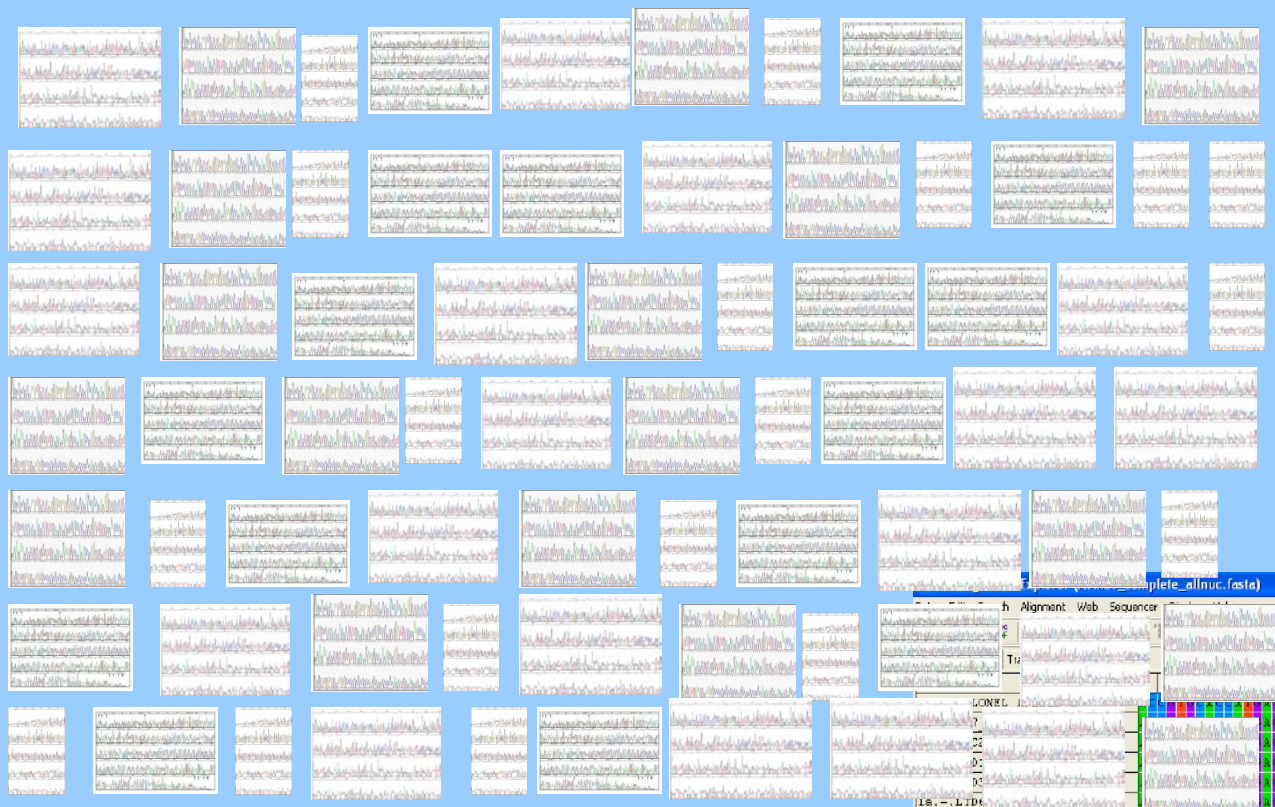
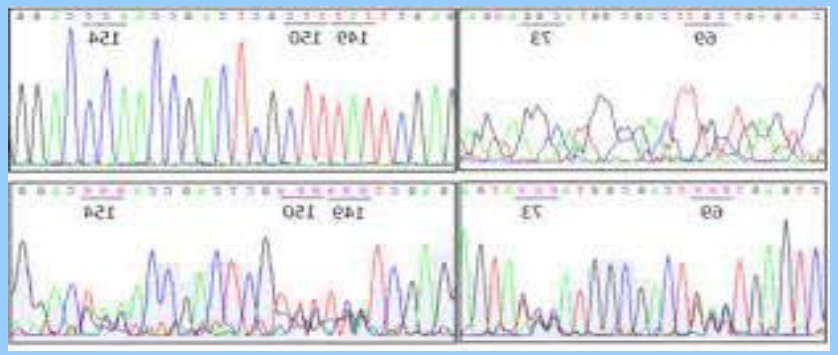
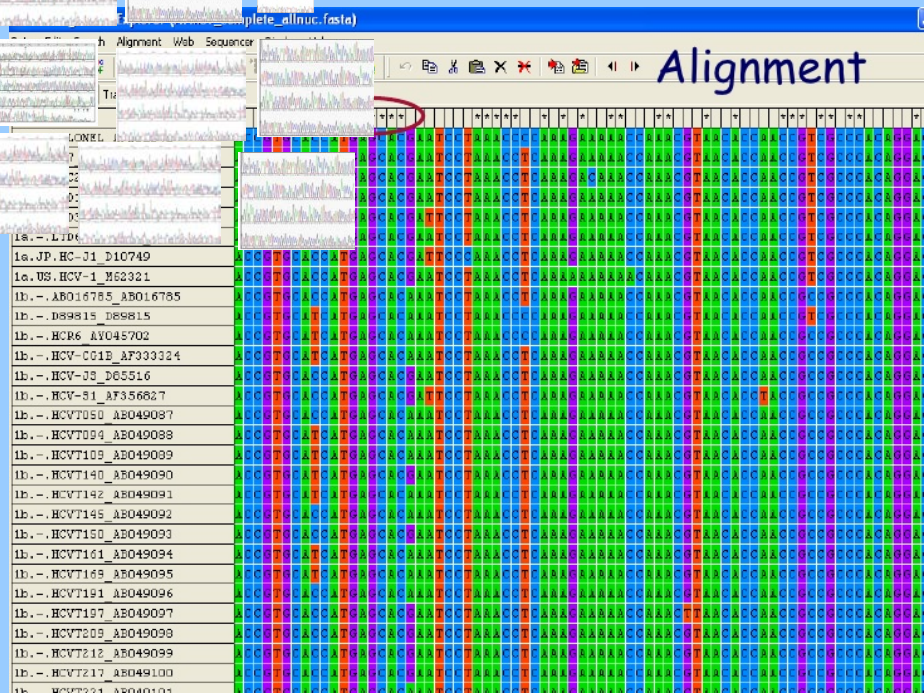
Forward strand



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1b.-.HCV-C01B_AF333324
1b.-.HCV-J8_D65516
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1b.-.HCVT145_AB049092
1b.-.HCVT150_AB049093
1b.-.HCVT161_AB049094
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1b.-.HCVT191_AB049096
1b.-.HCVT197_AB049097
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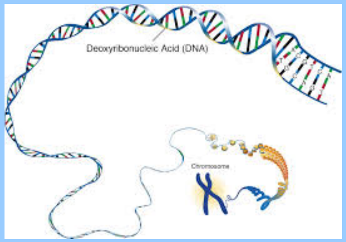
1b.-.HCVT217_AB049100

NEXT GENERATION SEQUENCING (NGS)



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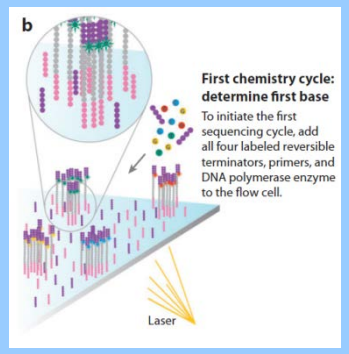
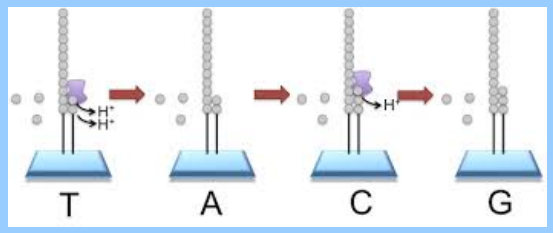
ILLUMINA



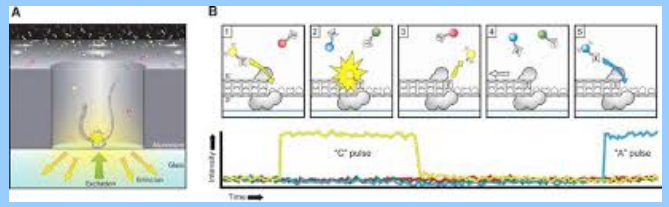
75-150 bases

PGM



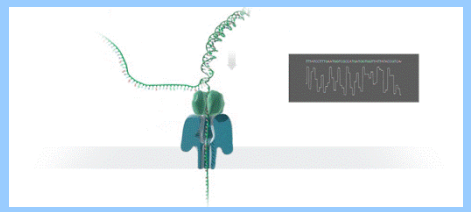
150-400 bases

PACIFIC B.

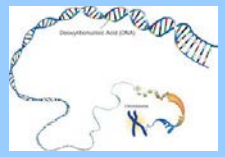


4000 bases

OXFORD NANOPORE



10000-20000 bases



CAPTURA
Gen
Panel
EXOMA

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+SRR566546.971 HWUSI-EAS1673_11067_FC7070M:4:1:2374:1108 length=50
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```

fastq, fq
Exoma 5-10 G

Genoma 500 G

ANALISIS BIOINFORMATICO

ENSAMBLAJE

(de novo)

ALINEAMIENTO

(genoma de referencia)

bioinformática

PIPELINE

FASTQ,FQ

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```

1) ALINEACION CON GENOMA DE REFERENCIA → SAM, BAM (BWA, BOWTIE, NOVOALING, SOAP,…)

```
1:497:R:-272+13M17D24M 113 1 497 37 37M 15 100338662 0 CGGGTCTGACCTGAGGAGAACTGTGCTCCGCCTTCAG 0;==-==9;>>>>>=>>>>>>>>>
19:20389:F:275+18M2D19M 147 1 17919 0 18M2D19M = 17644 -314 GTAGTACCAACTGTAAGTCCTTATCTTCATACTTTGT ;44999;499<8<8<<<8
9:21597+10M2I25M:R:-209 83 1 21678 0 8M2I27M = 21469 -244 CACCACATCACATATACCAAGCCTGGCTGTGTCTTCT <;9<<5><<<<><<<><><<>

. . .
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2) VARIANT CALL → VCF (GATK, FREEBAYES,…)

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20 2300608 rs84825     C T . PASS . GT:GP      0/1:0.03,0.97,0

20 2301308 rs84823     T G . PASS . GT:PL      1/1:10,5,0
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3) ANOTACION (ANNOVAR, KGGSEQ)

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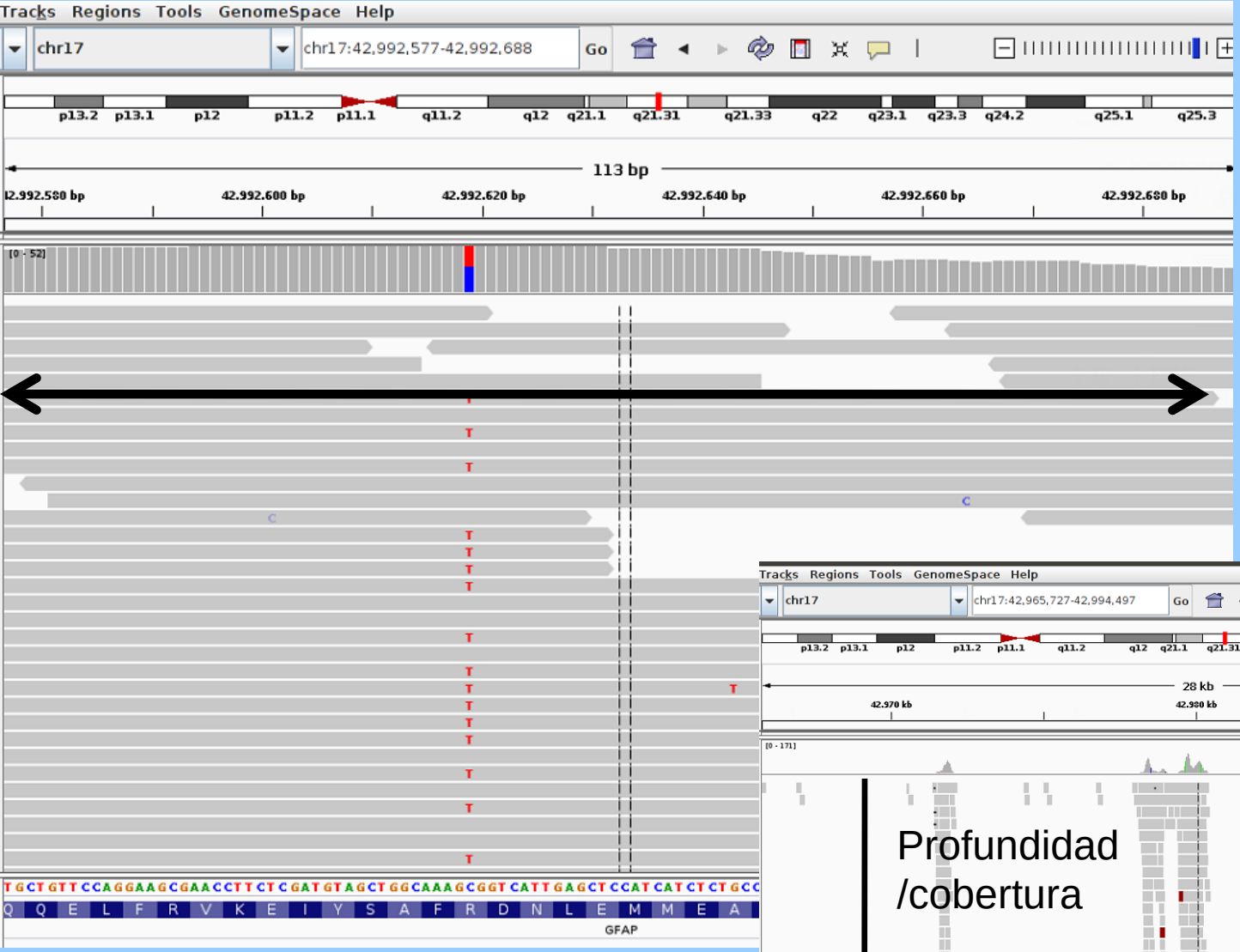
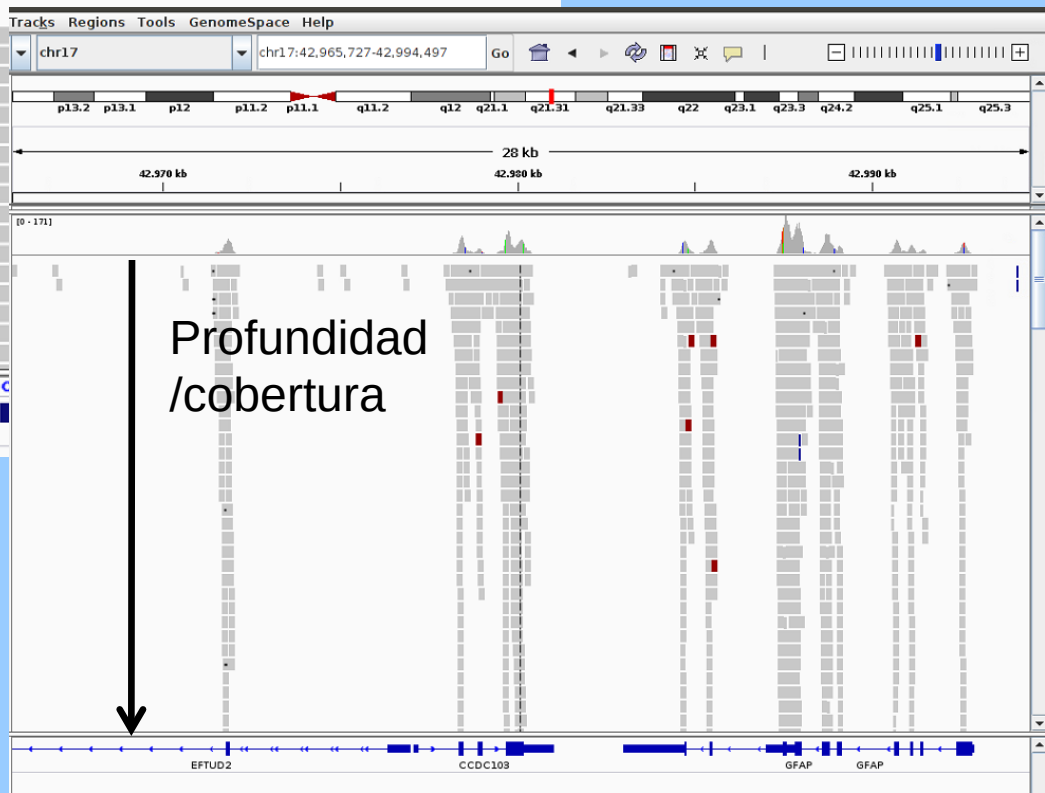
3	1	1404001	1404001	G	T	UTR3	ATAD3C	NM_001039211:c.*91G>T	.	.	1p36.33	Score=0.9(
4	1	5935162	5935162	A	T	splicing	NPHP4	NM_015102:exon22:c.2818-2T>A	.	.	1p36.31	.
5	1	162736463	162736463	C	T	intronic	DDR2	.	.	.	1q23.3	.
6	1	84875173	84875173	C	T	intronic	DNASE2B	.	.	.	1p31.1	.
7	1	13211293	13211294	TC	-	intergenic	HNRNPCP5,PRAMEF3	dist=26967;dist=116902	.	.	1p36.21	Score=0.9
8	1	11403596	11403596	-	AT	intergenic	UBIAD1,PTCHD2	dist=55105;dist=135699	.	.	1p36.22	.
9	1	105492231	105492231	A	ATAAA	intergenic	LOC100129138,NONE	dist=872538;dist=NONE	.	.	1p21.1	.
10	1	67705958	67705958	G	A	exonic	IL23R	.	nonsynonymous SNV	IL23R:NM_1447	1p31.3	.
11	2	234183368	234183368	A	G	exonic	ATG16L1	.	nonsynonymous SNV	ATG16L1:NM_1(	2q37.1	.
12	16	50745926	50745926	C	T	exonic	NOD2	.	nonsynonymous SNV	NOD2:NM_022(	16q12.1	.





genoma/exoma/panel

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GGAAACACTACTTAGGCTTATAAGATCNGGTTGCGG
+
ababbaaabaaaaa`]`ba`]`aaaaYD\\_a``XT
@IRIS:7:1:17:1757#0/1
TTTTCTCGACGATTTCCACTCCTGGTCNACGAATCC
+
aaaaaa``aaa`aaaa_^a```]][Z[DY^XYV^_Y
@IRIS:7:1:17:1479#0/1
CATATTGTAGGGTGGATCTCGAAAGATATGAAAGAT
+
abaaaaa`a```^aaaaa`_]aaa`aaa__a_X]``
.........
.........
```

PIPELINE

1) ALINEACION CON GENOMA DE REFERENCIA → SAM, BAM (BWA, BOWTIE, NOVO

```
1:497:R:-272+13M17D24M 113 1 497 37 37M 15 100338662 0 CGGGTCTGACCTG
==9;>>>>>=>>>>>>>>>>>=>>>>>>>>>>>>>>
19:20389:F:275+18M2D19M 147 1 17919 0 18M2D19M = 17644 -314 GTAGTACC
;44999;499<8<8<<<8<<><<<<<><7<;<<<<<<
9:21597+10M2I25M:R:-209 83 1 21678 0 8M2I27M = 21469 -244 CACCACATCA
<;9<<5><<<<><<<><><><><9>
...
```

2) VARIANT CALL → VCF (GATK, FREEBAYES,...

```
#CHROM POS ID  REF ALT QUAL FILTER INFO
20 1291018 rs11449            G A . PASS . GT                0/0 0/1
20 2300608 rs84825            C T . PASS . GT:GP             0/1:0.03,
20 2301308 rs84823            T G . PASS . GT:PL             1/1:10,5,
....
```

3) ANOTACION (ANNOVAR, KGGSEQ)

	Chr	Start	End	Ref	Alt	Func.refGene	Gene.refGene	GeneDetail.refGene	ExonicFunc.refGene	AAChange.refGe	cytoBand	genomicSu
2	1	948921	948921	T	C	UTR5	ISG15	NM_005101:c.-33T>C	.	.	1p36.33	.
3	1	1404001	1404001	G	T	UTR3	ATAD3C	NM_001039211:c.*91G>T	.	.	1p36.33	Score=0.9
4	1	5935162	5935162	A	T	splicing	NPHP4	NM_015102:exon22:c.2818-2T>A	.	.	1p36.31	.
5	1	162736463	162736463	C	T	intergenic	DDR2	.	.	.	1q23.3	.
6	1	84875173	84875173	C	T	intronic	DNASE2B	.	.	.	1p31.1	.
7	1	13211293	13211294	TC	-	intergenic	HNRNPCP5,PRAMEF3	dist=26967;dist=116902	.	.	1p36.21	Score=0.9
8	1	11403596	11403596	-	AT	intergenic	UBIAD1,PTCHD2	dist=55105;dist=135699	.	.	1p36.22	.
9	1	105492231	105492231	A	ATAAA	intergenic	LOC100129138,NONE	dist=872538;dist=NONE	.	.	1p21.1	.
10	1	67705958	67705958	G	A	exonic	IL23R	.	nonsynonymous SNV	IL23R:NM_1447	1p31.3	.
11	2	234183368	234183368	A	G	exonic	ATG16L1	.	nonsynonymous SNV	ATG16L1:NM_1	2q37.1	.
12	16	50745926	50745926	C	T	exonic	NOD2	.	nonsynonymous SNV	NOD2:NM_022	16q12.1	.

FILTRADO

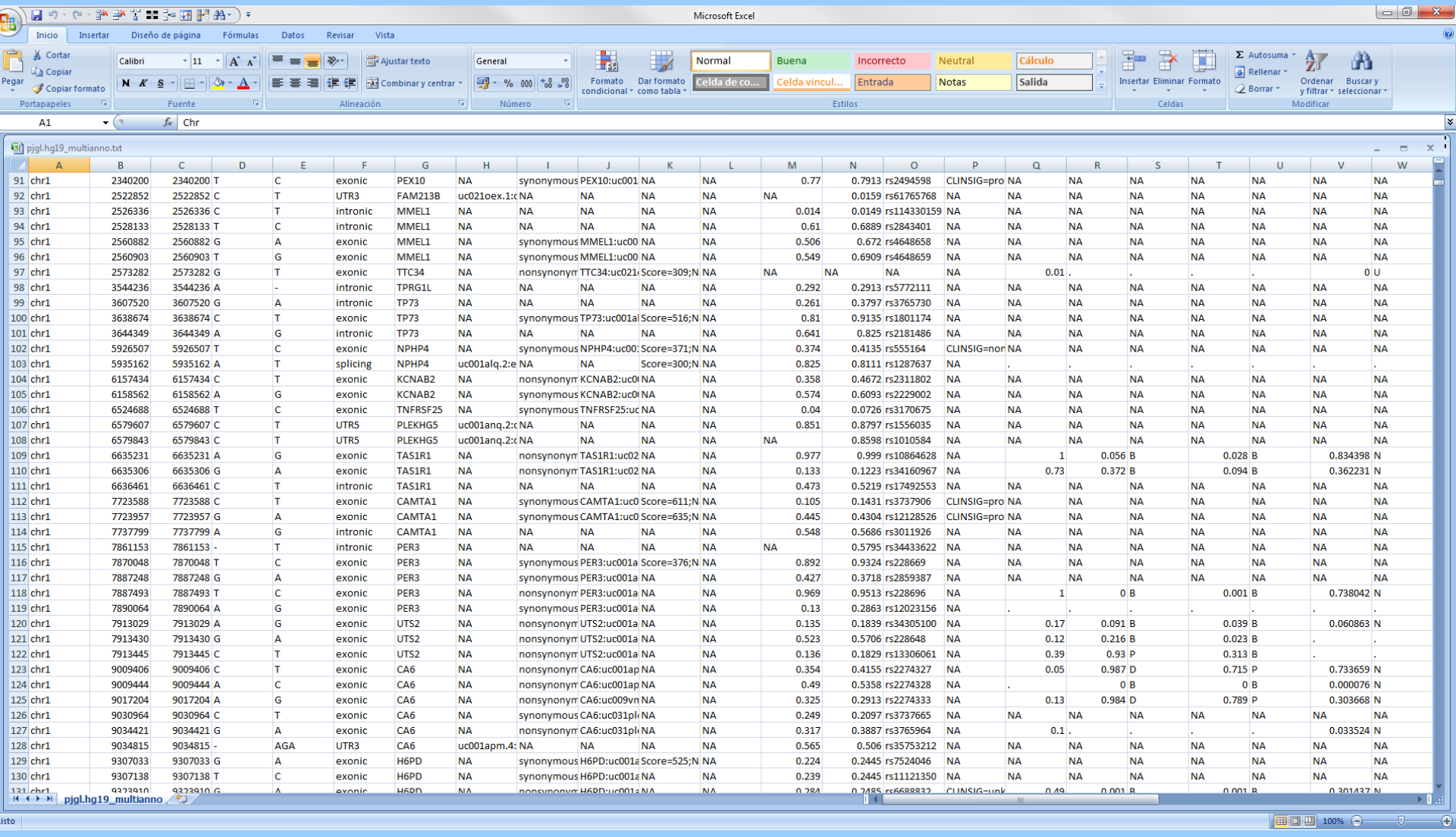
30000 VARIANTES TOTALES

1000 VARIABLES INFRECUENTES

200 FILTRADO ESTRUCTURAL (MISSENSE, SPLICING)

8 GENES CANDIDATOS

1-3 PATRON HERENCIA/TRIOS/SEGREGACION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pjgl.hg19_multianno.txt
pjgl.hg19_multianno
100%

Microsoft Excel

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Insertar | Eliminar | Formato — Celdas

Autosuma | Rellenar | Borrar | Ordenar y filtrar | Buscar y seleccionar — Modificar

H1

=BUSCARV(G1;$A$40700:$A$40900;1;0)

iam.hg19_multianno.txt

	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	
55	chr1	888659	888659	T	C	exonic	NOC2L	#N/A	NA	nonsynonym	NOC2L:uc001	NA	NA	0.935	0.9533	rs3748597	NA	1	
56	chr1	889158	889158	G	C	intronic	NOC2L	#N/A	NA	NA	NA	NA	NA	0.927	0.9523	rs13303056	NA	NA	NA
57	chr1	889159	889159	A	C	intronic	NOC2L	#N/A	NA	NA	NA	NA	NA	0.927	0.9523	rs13302945	NA	NA	NA
58	chr1	897325	897325	G	C	exonic	KLHL17	#N/A	NA	synonymous	KLHL17:uc00	NA	NA	0.907	0.9433	rs4970441	NA	NA	NA
59	chr1	899928	899928	G	C	intronic	KLHL17	#N/A	NA	NA	NA	NA	NA	0.807	0.9414	rs6677386	NA	NA	NA
60	chr1	906302	906302	C	T	exonic	PLEKHN1	#N/A	NA	synonymous	PLEKHN1:uc(	Score=369;N	NA	4.85E-03	0.0089	rs41300090	NA	NA	NA
61	chr1	909238	909238	G	C	exonic	PLEKHN1	#N/A	NA	nonsynonym	PLEKHN1:uc(	NA	NA	0.665	0.6183	rs3829740	NA	0.28	
62	chr1	910394	910394	C	T	UTR3	PLEKHN1	#N/A	uc001acd.3:c	NA	NA	NA	NA	NA	0.3211	rs28477686	NA	NA	NA
63	chr1	910409	910409	T	C	UTR3	PLEKHN1	#N/A	uc001acd.3:c	NA	NA	NA	NA	NA	0.1272	rs28536514	NA	NA	NA
64	chr1	935222	935222	C	A	exonic	HES4	#N/A	NA	nonsynonym	HES4:uc010n	NA	NA	0.57	0.5895	rs2298214	NA	1	
65	chr1	948846	948846	-	A	upstream	ISG15	#N/A	NA	NA	NA	NA	NA	NA	0.9791	rs3841266	NA	NA	NA
66	chr1	948870	948870	C	G	UTR5	ISG15	#N/A	uc001acj.4:c.	NA	NA	NA	NA	NA	0.9573	rs4615788	NA	NA	NA
67	chr1	948921	948921	T	C	UTR5	ISG15	#N/A	uc001acj.4:c.	NA	NA	NA	NA	0.941	0.9573	rs15842	NA	NA	NA
68	chr1	949608	949608	G	A	exonic	ISG15	#N/A	NA	nonsynonym	ISG15:uc001a	NA	NA	0.369	0.3926	rs1921	NA	0	
69	chr1	949654	949654	A	G	exonic	ISG15	#N/A	NA	synonymous	ISG15:uc001a	NA	NA	0.912	0.9453	rs8997	NA	NA	NA
70	chr1	978604	978605	CT	-	intronic	AGRN	#N/A	NA	NA	NA	NA	NA	0.497	0.5487	rs35881187	NA	NA	NA
71	chr1	981931	981931	A	G	exonic	AGRN	#N/A	NA	synonymous	AGRN:uc001	NA	NA	0.872	0.9125	rs2465128	CLINSIG=non	NA	NA
72	chr1	982941	982941	T	C	intronic	AGRN	#N/A	NA	NA	NA	NA	NA	0.883	0.9175	rs3128102	NA	NA	NA
73	chr1	982994	982994	T	C	exonic	AGRN	#N/A	NA	synonymous	AGRN:uc001	Score=482;N	NA	0.898	0.9205	rs10267	CLINSIG=non	NA	NA
74	chr1	984302	984302	T	C	exonic	AGRN	#N/A	NA	synonymous	AGRN:uc001	NA	NA	0.501	0.5636	rs9442391	CLINSIG=non	NA	NA
75	chr1	985266	985266	C	T	intronic	AGRN	#N/A	NA	NA	NA	NA	NA	0.535	0.5457	rs2275813	NA	NA	NA
76	chr1	987200	987200	C	T	intronic	AGRN	#N/A	NA	NA	NA	NA	NA	0.874	0.9145	rs9803031	CLINSIG=non	NA	NA
77	chr1	990280	990280	C	T	exonic	AGRN	#N/A	NA	synonymous	AGRN:uc001	Score=582;N	NA	0.574	0.6779	rs4275402	CLINSIG=non	NA	NA
78	chr1	990517	990517	C	T	UTR3	AGRN	#N/A	uc001ack.2:c	NA	NA	NA	NA	NA	0.8877	rs2710872	NA	NA	NA
79	chr1	1158631	1158631	A	G	exonic	SDF4	#N/A	NA	synonymous	SDF4:uc001a	NA	NA	0.916	0.8608	rs6603781	NA	.	.
80	chr1	1226512	1226512	G	A	exonic	SCNN1D	#N/A	NA	nonsynonym	SCNN1D:uc0	NA	NA	0.015	0.0179	rs114257304	NA	0.07	
81	chr1	1246004	1246004	A	G	exonic	PUSL1	#N/A	NA	nonsynonym	PUSL1:uc009	NA	NA	0.96	0.9672	rs2296474	NA	NA	NA
82	chr1	1247494	1247494	T	C	exonic	CPSF3L	#N/A	NA	synonymous	CPSF3L:uc00	NA	NA	0.543	0.8121	rs12103	NA	NA	NA
83	chr1	1249187	1249187	G	A	exonic	CPSF3L	#N/A	NA	synonymous	CPSF3L:uc00	Score=620;N	NA	0.58	0.8052	rs12142199	NA	NA	NA
84	chr1	1254841	1254841	C	G	exonic	CPSF3L	#N/A	NA	synonymous	CPSF3L:uc00	Score=625;N	NA	0.847	0.9205	rs10907179	NA	NA	NA
85	chr1	1262966	1262966	C	T	exonic	GLTPD1	#N/A	NA	synonymous	GLTPD1:uc00	Score=496;N	NA	0.857	0.9155	rs307349	NA	NA	NA
86	chr1	1263144	1263144	G	A	UTR3	GLTPD1	#N/A	uc001aeo.3:c	NA	NA	NA	NA	0.889	0.9235	rs307350	NA	NA	NA
87	chr1	1263362	1263362	G	A	UTR3	GLTPD1	#N/A	uc001aeo.3:c	NA	NA	NA	NA	NA	0.9205	rs4970433	NA	NA	NA
88	chr1	1263457	1263457	T	C	UTR3	GLTPD1	#N/A	uc001aeo.3:c	NA	NA	NA	NA	NA	0.9662	rs307351	NA	NA	NA
89	chr1	1269554	1269554	T	C	exonic	TAS1R3	#N/A	NA	nonsynonym	TAS1R3:uc01	NA	NA	0.957	0.9662	rs307377	NA	0.33	
90	chr1	1277533	1277533	T	C	exonic	DVL1	#N/A	NA	synonymous	DVL1:uc001a	Score=513;N	NA	0.995	0.994	rs307362	NA	NA	NA
91	chr1	1288823	1288823	A	G	UTR3	MXRA8	#N/A	uc001aew.3:c	NA	NA	NA	NA	NA	0.9205	rs2296471	NA	NA	NA
92	chr1	1296691	1296691	C	G	UTR5	MXRA8	#N/A	uc001afa.3:c	NA	NA	NA	NA	0.938	0.9662	rs2765025	NA	NA	NA
93	chr1	1296818	1296818	C	G	UTR5	MXRA8	#N/A	uc001afa.3:c	NA	NA	NA	NA	NA	0.9662	rs2765024	NA	NA	NA
94	chr1	1310074	1310074	C	G	intronic	AURKAIP1	#N/A	NA	NA	NA	NA	NA	0.874	0.9364	rs2765035	NA	NA	NA

iam.hg19_multianno

Pegar | Copiar | Copiar formato — Portapapeles

Arial | 10 — Fuente

A1

ACT

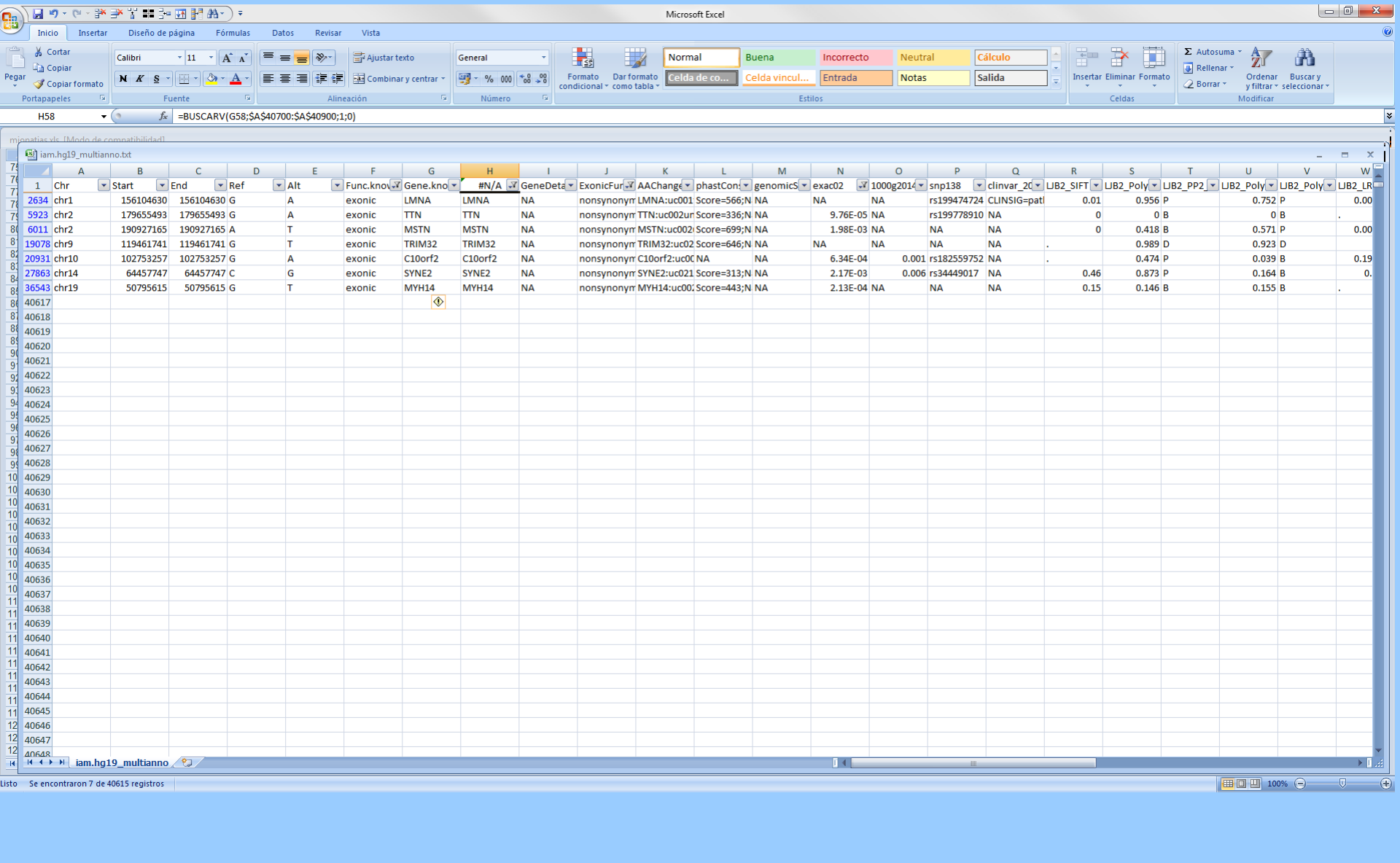
miopatias.xls [Modo de compatibilidad]

	A
40	KLHL40
41	KLHL41
42	KLHL9
43	LAMA2
44	LAMP2
45	LARGE
46	LDB3
47	LMNA
48	MATR3
49	MEGF10
50	MSTN
51	MTM1
52	MYBPC1
53	MYBPC3
54	MYF6
55	MYH14
56	MYH2
57	MYH7
58	MYOT
59	MYOT
60	NEB
61	OPA1
62	PABPN1
63	PLEC
64	POLG
65	POLG2
66	POMGNT1
67	POMT1
68	POMT2
69	PTPLA
70	PTRF
71	PUS1
72	RRM2B
73	RYR1
74	SEPN1
75	SGCA
76	SGCB
77	SGCD
78	SGCG
79	SIL1
80	SMCHD1
81	STAC3
82	STIM1
83	SUCLA2
84	SYNE1
85	SYNE2
86	TCAP
87	TIA1

distrofias | miasteniacongenita

Seleccione el destino y presione ENTRAR o elija Pega

964	chr1	26140573	26140573	C	A	exonic	SEPN1	SEPN1	NA	unknown	UNKNOWN	NA
1313	chr1	46655158	46655158	T	C	exonic	POMGNT1	POMGNT1	NA	nonsynonym	POMGNT1:u	Score
2634	chr1	156104630	156104630	G	A	exonic	LMNA	LMNA	NA	nonsynonym	LMNA:uc001	Score
3717	chr1	229566998	229566998	C	T	UTR3	ACTA1	ACTA1	uc001htm.3:c	NA	NA	NA
3718	chr1	229567660	229567660	-	G	intronic	ACTA1	ACTA1	NA	NA	NA	NA
3719	chr1	229567663	229567663	C	G	intronic	ACTA1	ACTA1	NA	NA	NA	NA
3720	chr1	229568632	229568632	A	G	intronic	ACTA1	ACTA1	NA	NA	NA	NA
3721	chr1	229568637	229568637	C	G	intronic	ACTA1	ACTA1	NA	NA	NA	NA
4806	chr2	71742736	71742736	C	T	intronic	DYSF	DYSF	NA	NA	NA	NA
4807	chr2	71747899	71747899	G	A	intronic	DYSF	DYSF	NA	NA	NA	NA
4808	chr2	71753487	71753487	C	T	intronic	DYSF	DYSF	NA	NA	NA	NA
4809	chr2	71762232	71762232	C	T	intronic	DYSF	DYSF	NA	NA	NA	NA
4810	chr2	71780215	71780215	T	C	exonic	DYSF	DYSF	NA	synonymous	DYSF:uc002si	Score
4811	chr2	71783217	71783217	C	T	intronic	DYSF	DYSF	NA	NA	NA	NA
4812	chr2	71795152	71795152	A	T	exonic	DYSF	DYSF	NA	synonymous	DYSF:uc002si	Score
4813	chr2	71797712	71797712	G	A	intronic	DYSF	DYSF	NA	NA	NA	NA
4814	chr2	71906171	71906171	T	C	intronic	DYSF	DYSF	NA	NA	NA	NA
4815	chr2	71906278	71906278	A	C	exonic	DYSF	DYSF	NA	synonymous	DYSF:uc010y	NA
4816	chr2	71909822	71909822	C	T	intronic	DYSF	DYSF	NA	NA	NA	NA
5303	chr2	127819777	127819777	C	T	intronic	BIN1	BIN1	NA	NA	NA	NA
5603	chr2	152346979	152346979	T	C	exonic	NEB	NEB	NA	nonsynonym	NEB:uc010zb	Score
5604	chr2	152349026	152349026	G	A	intronic	NEB	NEB	NA	NA	NA	NA
5605	chr2	152352843	152352843	C	G	exonic	NEB	NEB	NA	nonsynonym	NEB:uc010zb	Score
5606	chr2	152382454	152382454	C	G	intronic	NEB	NEB	NA	NA	NA	NA
5607	chr2	152387553	152387553	T	C	exonic	NEB	NEB	NA	synonymous	NEB:uc002tx	NA
5608	chr2	152388416	152388416	T	-	UTR5	NEB	NEB	uc002txt.4:c.	NA	NA	Score
5609	chr2	152422076	152422076	C	G	exonic	NEB	NEB	NA	nonsynonym	NEB:uc002tx	Score
5610	chr2	152422087	152422087	A	G	exonic	NEB	NEB	NA	synonymous	NEB:uc002tx	Score
5611	chr2	152424933	152424933	-	A	splicing	NEB	NEB	uc002txr.3:e:	NA	NA	Score
5612	chr2	152432886	152432886	G	A	intronic	NEB	NEB	NA	NA	NA	NA
5613	chr2	152436012	152436012	T	G	exonic	NEB	NEB	NA	nonsynonym	NEB:uc002tx	Score
5614	chr2	152438107	152438107	A	G	intronic	NEB	NEB	NA	NA	NA	NA
5615	chr2	152447845	152447845	-	C	intronic	NEB	NEB	NA	NA	NA	NA
5616	chr2	152448640	152448640	T	C	exonic	NEB	NEB	NA	nonsynonym	NEB:uc002tx	Score
5617	chr2	152474001	152474001	A	G	intronic	NEB	NEB	NA	NA	NA	NA
5618	chr2	152490219	152490219	A	G	exonic	NEB	NEB	NA	synonymous	NEB:uc002tx	NA



1	Chr	Start	End	Ref	Alt	Func.kno	Gene.kno	#N/A	GeneDeta	ExonicFur	AAChange	phastCon	genomicS	exac02	1000g2014	snp138	clinvar_20	LJB2_SIFT	LJB2_Poly	LJB2_PP2	LJB2_Poly	LJB2_Poly	LJB2_LR
2634	chr1	156104630	156104630	G	A	exonic	LMNA	LMNA	NA	nonsynonym	LMNA:uc001	Score=566;N	NA	NA	NA	rs199474724	CLINSIG=pat	0.01	0.956	P	0.752	P	0.00
5923	chr2	179655493	179655493	G	A	exonic	TTN	TTN	NA	nonsynonym	TTN:uc002ur	Score=336;N	NA	9.76E-05	NA	rs199778910	NA	0	0	B	0	B	.
6011	chr2	190927165	190927165	A	T	exonic	MSTN	MSTN	NA	nonsynonym	MSTN:uc002	Score=699;N	NA	1.98E-03	NA	NA	NA	0	0.418	B	0.571	P	0.00
19078	chr9	119461741	119461741	G	T	exonic	TRIM32	TRIM32	NA	nonsynonym	TRIM32:uc02	Score=646;N	NA	NA	NA	NA	NA	.	0.989	D	0.923	D	
20931	chr10	102753257	102753257	G	A	exonic	C10orf2	C10orf2	NA	nonsynonym	C10orf2:uc0	NA	NA	6.34E-04	0.001	rs182559752	NA	.	0.474	P	0.039	B	0.19
27863	chr14	64457747	64457747	C	G	exonic	SYNE2	SYNE2	NA	nonsynonym	SYNE2:uc021	Score=313;N	NA	2.17E-03	0.006	rs34449017	NA	0.46	0.873	P	0.164	B	0.
36543	chr19	50795615	50795615	G	T	exonic	MYH14	MYH14	NA	nonsynonym	MYH14:uc00	Score=443;N	NA	2.13E-04	NA	NA	NA	0.15	0.146	B	0.155	B	.


aje@aje-OptiPlex-990: ~/Public/kggseq
Es
8:50
aje@aje-OptiPlex-990:~/Public$ cd kggseq
aje@aje-OptiPlex-990:~/Public/kggseq$ java -Xms256m -Xmx1300m -jar kggseq.jar --no-resource-check --no-lib-check --buildver hg19 --vcf-file /home/aje/Public/men/controlesHC.vcf
--ped-file /home/aje/Public/men/control.ped --db-gene refgene --gene-feature-in 0,1,2,3,4,5,7 --splicing 15 --genotype-filter 1,2 --db-filter hg19_1kg201204,exac --db-score db
nsfp --excel --gene -var- filter 5 --filter-model case-unique --superdup-filter --genome-annot --omim-annot --genes-in ACTA1,ANO5,ANO5,ADCY6,ATP2A1,ABHD5,ACVR1,CAPN3,CHKB,AG
RN,CACNA1A,ACAD9,ANO5,CAV3,COL6A1,ALG2,CACNA1S,ACADL,BAG3,DAG1,COL6A2,ALG14,CAV3,ACADM,BIN1,DES,COL6A3,CHAT,CLCN1,ACADS,C10orf2,DNAJB6,DMD,CHRNA1,HINT1,ACADVL,CAV3,DPM3,DNM2,CH
RNB1,HSPG2,AGL,CCDC78,DYSF,DPM2,CHRND,KCNA1,AMPD1,CFL2,FKRP,DYSF,CHRNE,KCNE3,CPT1B,CNTN1,FKTN,EMD,CHRNG,KCNJ18,CPT2,COL12A1,GMPPB,FHL1,CNTNAP1,SCN4A,ENO3,COL6A1,LMNA,FKRP,COLQ,
ETFA,COL6A2,MYOT,FKTN,DOK7,ETFB,COL6A3,PLEC,GMPPB,DPAGT1,ETFDH,CRYAB,POMGNT1,ISPD,ECEL1,GAA,DES,POMT1,ITGA7,ERBB3,GBE1,DNM2,POMT2,LAMA2,FBN2,GYG1,DYSF,SGCA,LARGE,GFPT1,GYS1,FHL
1,SGCB,LMNA,GLE1,HADH,FKBP14,SGCD,PABPN1,IBA57,HADHA,FLNC,SGCG,POMGNT1,LAMB2,HADHB,GNE,TCAP,POMT1,MUSK,LDHA,HNRNPA1,TNPO3,POMT2,MYBPC1,LPIN1,HNRNPA2B1,TRAPPC11,PTRF,MYH3,PDHA1,
ISCU,TRIM32,SEPN1,MYH8,PFKM,KBTBD13,TTN,SMCHD1,PIEZO2,PGAM2,KLHL40,SYNE1,PIP5K1C,PGK1,KLHL41,SYNE2,PLEC,PGM1,KLHL9,TCAP,PLOD2,PHKA1,LAMP2,TMEM43,PREPL,PNPLA2,LDB3,TTN,RAPSN,POL
G2,MATR3,SCN4A,PRKAG2,MEGF10,SYNE1,PUS1,MSTN,TNNI2,PYGM,MTM1,TNNT3,RRM2B,MYBPC1,TPM2,SLC16A1,MYBPC3,ZC4H2,SLC22A5,MYF6,SLC25A20,MYH2,TAZ,MYH7,YARS2,MYH14,MYOT,NEB,OPA1,PABPN1,P
LEC,POLG,POLG2,PTPLA,PUS1,RRM2B,RYR1,SEPN1,SIL1,STAC3,STIM1,SUCLA2,TIA1,TK2,TNNT1,TPM2,TPM3,TRIM32,TTN,VCP,VMA21,YARS2

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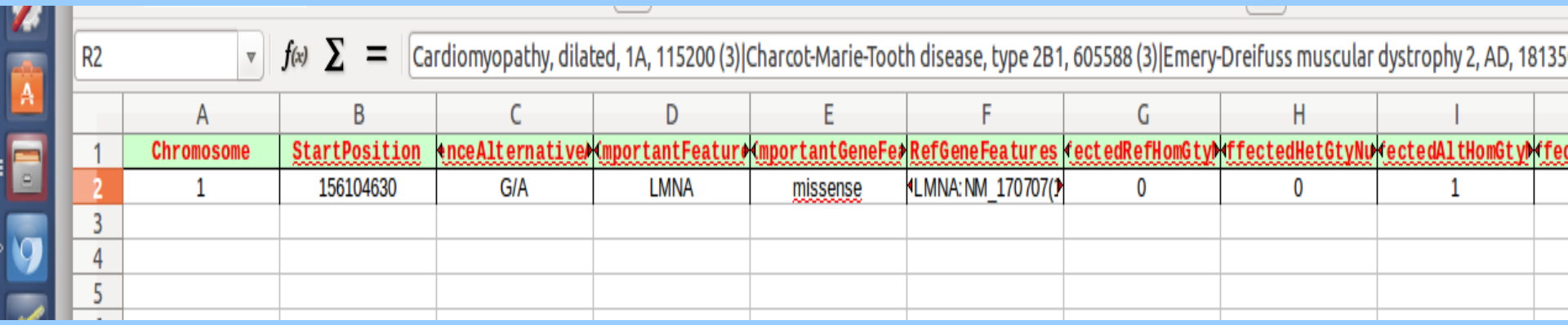
--filter-model case-unique
--genes-in ACTA1,ANO5,ADCY6,ATP2A1,ABHD5,ACVR1,CAPN3,CHKB,AGRN,CACNA1A,ACAD9,CAV3,COL6A1,ALG2,CACNA1S,ACADL,BAG3,DAG1,COL6A2,ALG14,ACADM,BIN1,DES,COL6A3,CHAT,CLCN1,ACADS,C10orf
2,DNAJB6,DMD,CHRNA1,HINT1,ACADVL,DPM3,DNM2,CHRNB1,HSPG2,AGL,CCDC78,DYSF,DPM2,CHRND,KCNA1,AMPD1,CFL2,FKRP,CHRNE,KCNE3,CPT1B,CNTN1,FKTN,EMD,CHRNG,KCNJ18,CPT2,COL12A1,GMPPB,FHL1,C
NTNAP1,SCN4A,ENO3,LMNA,COLQ,ETFA,MYOT,DOK7,ETFB,PLEC,DPAGT1,ETFDH,CRYAB,POMGNT1,ISPD,ECEL1,GAA,POMT1,ITGA7,ERBB3,GBE1,POMT2,LAMA2,FBN2,GYG1,SGCA,LARGE,GFPT1,GYS1,SGCB,GLE1,HADH
,FKBP14,SGCD,PABPN1,IBA57,HADHA,FLNC,SGCG,LAMB2,HADHB,GNE,TCAP,MUSK,LDHA,HNRNPA1,TNPO3,MYBPC1,LPIN1,HNRNPA2B1,TRAPPC11,PTRF,MYH3,PDHA1,ISCU,TRIM32,SEPN1,MYH8,PFKM,KBTBD13,TTN,S
MCHD1,PIEZO2,PGAM2,KLHL40,SYNE1,PIP5K1C,PGK1,KLHL41,SYNE2,PGM1,KLHL9,PLOD2,PHKA1,LAMP2,TMEM43,PREPL,PNPLA2,LDB3,RAPSN,POLG2,MATR3,PRKAG2,MEGF10,PUS1,MSTN,TNNI2,PYGM,MTM1,TNNT3,
RRM2B,TPM2,SLC16A1,MYBPC3,ZC4H2,SLC22A5,MYF6,SLC25A20,MYH2,TAZ,MYH7,YARS2,MYH14,NEB,OPA1,POLG,PTPLA,RYR1,SIL1,STAC3,STIM1,SUCLA2,TIA1,TK2,TNNT1,TPM3,VCP,VMA21
---genome-annot
--omim-annot
--superdup-filter
--db-score dbnsfp
--mendel-causing-predict
INFO  2015-03-25 08:50:53 - Encoding pedigree file /home/aje/Public/men/control.ped ...
INFO  2015-03-25 08:50:53 - Reading variants in /home/aje/Public/men/controlesHC.vcf
INFO  2015-03-25 08:51:04 -
6867635 missing genotypes are ignored
6893 genotypes are ignored due to low depth at the site with a genotype <4
142477 genotypes are ignored due to low quality of specific genotyping quality <10.0
37344 genotypes are ignored because the fraction of the reads carrying alternative allele >= 0.05 at a reference-allele homozygous genotype and that is <= 0.25 at a heterozygou
s genotype and that is <= 0.75 at an alternative-allele homozygous genotype
114708 genotypes are ignored because their second smallest Phred-scaled likelihoods (PL) are < 20.

299723 variants are ignored due to low sequencing quality (<50.0)

765214 variant-lines (76493 indels) are scanned; and 465491 variants of 17 individual(s) are valid in /home/aje/Public/men/controlesHC.vcf
INFO  2015-03-25 08:51:04 - 20041 variants are ignored by genotype-based hard-filtering;
445450 variant(s) are left after filtration according to inheritance mode at genotypes, 1,2!
INFO  2015-03-25 08:51:04 - 66 variant(s) are left after filtration according to case-unique!
Parsed variants INFO  2015-03-25 08:51:25 - 43 variant(s) exist in 1kg201204, which contains 39706714 effective variants.
Parsed variants INFO  2015-03-25 08:51:30 - 35 variant(s) exist in exac, which contains 10205998 effective variants.
INFO  2015-03-25 08:51:30 - 26 variant(s) with minor allele frequency [0, 0.01) in the reference datasets above are left!
INFO  2015-03-25 08:51:33 - 52259 transcripts of 25953 genes have been read in the dataset kggseq_hg19_refGene.txt.gz!
INFO  2015-03-25 08:51:34 -
0.frameshift: 0 (0%)
1.nonframeshift: 0 (0%)
2.stoploss: 0 (0%)
3.stopgain: 0 (0%)
4.missense: 7 (26.923%)
5.splicing: 0 (0%)
6.synonymous: 1 (3.846%)
7.exonic: 0 (0%)
8.5UTR: 0 (0%)
9.3UTR: 8 (30.769%)
10.intronic: 3 (11.538%)
11.upstream: 0 (0%)
12.downstream: 0 (0%)
13.ncRNA: 0 (0%)
14.intergenic: 0 (0%)
15.monomorphic: 0 (0%)
16.unknown: 7 (26.923%)

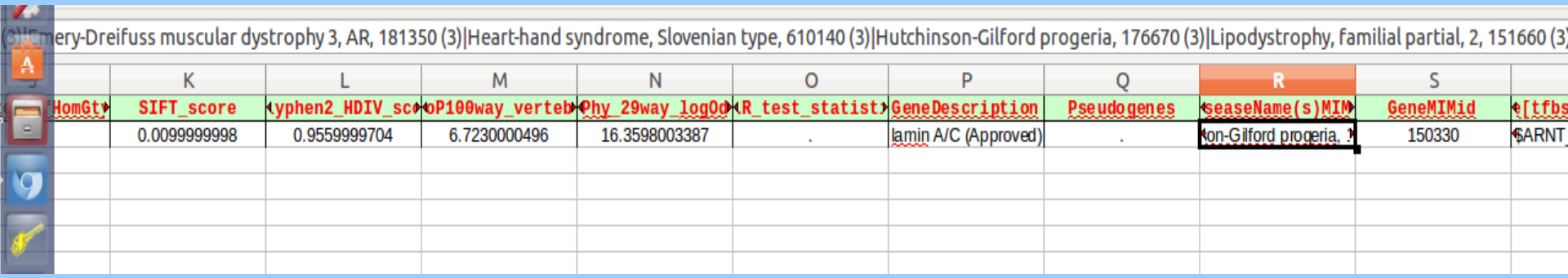
INFO  2015-03-25 08:51:34 - 7 variant(s) are left after filtering by gene features.
INFO  2015-03-25 08:51:34 - 1 variant(s) are left after filtering those outside specified genes!
INFO  2015-03-25 08:51:34 - 1 sequence variant(s) are highlighted by OMIM information!
INFO  2015-03-25 08:51:35 - 41160 reference super duplicate regions have been read!
INFO  2015-03-25 08:51:35 - 1 variant(s) are left after filtering those in super-duplicated regions registered in a data set genomicSuperDups table of UCSC!
Parsed variants
Parsed variants ords
Parsed variants INFO  2015-03-25 08:51:47 - 1 coding nonsynonymous variants among 1 are assigned functional prediction scores.
INFO  2015-03-25 08:51:47 - The number of predicted disease-causal and non-disease-causal variants are 1(#gene 1) and 0 among 1 variants on the genome according to the Logistic
 regression prediction model trained by ExoVar dataset (http://statgenpro.psychiatry.hku.hk/limx/kggseq/download/ExoVar.xls)
INFO  2015-03-25 08:51:50 - Final variants are saved in /home/aje/Public/kggseq/kggseq.flt.xlsx with Excel format.
INFO  2015-03-25 08:51:50 - 1 sequence variant(s) are left finally!
INFO  2015-03-25 08:51:50 - Elapsed time: 0 min. 56 sec.
The log information is save in ./kggseq.log
aje@aje-OptiPlex-990:~/Public/kggseq$
```



R2

Cardiomyopathy, dilated, 1A, 115200 (3)|Charcot-Marie-Tooth disease, type 2B1, 605588 (3)|Emery-Dreifuss muscular dystrophy 2, AD, 18135

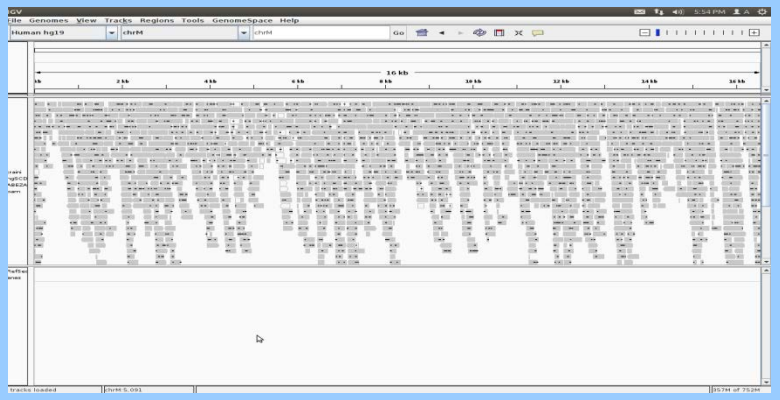
	A	B	C	D	E	F	G	H	I
1	Chromosome	StartPosition	nceAlternative	mportantFeatur	mportantGeneFe	RefGeneFeatures	ectedRefHomGty	ffectedHetGtyNu	ectedAltHomGty	ffe
2	1	156104630	G/A	LMNA	missense	LMNA:NM_170707(1	0	0	1	
3										
4										
5										



(3)Emery-Dreifuss muscular dystrophy 3, AR, 181350 (3)|Heart-hand syndrome, Slovenian type, 610140 (3)|Hutchinson-Gilford progeria, 176670 (3)|Lipodystrophy, familial partial, 2, 151660 (3)

	K	L	M	N	O	P	Q	R	S	
HomGt	SIFT_score	yphen2_HDIV_sc	oP100way_verteb	Phy_29way_logOd	R_test_statist	GeneDescription	Pseudogenes	seaseName(s)MIM	GeneMIMid	[tfbs
	0.0099999998	0.9559999704	6.7230000496	16.3598003387	.	lamin A/C (Approved)	.	lon-Gilford progeria, 1	150330	SARNT

Estudio de genética mitocondrial con NGS



Enfermedades mitocondriales:
-mutaciones puntuales
-deleciones, duplicaciones

Trastornos mitocondriales
S. Fatiga crónica
HiperCKemia
Intolerancia ejercicio

OPEN ACCESS Freely available online

PLOS | COMPUTATIONAL BIOLOGY

Next-Generation Sequencing of Human Mitochondrial Reference Genomes Uncovers High Heteroplasmy Frequency

Maria Ximena Sosa[1☯], I. K. Ashok Sivakumar[1,2☯], Samantha Maragh[1,3], Vamsi Veeramachaneni[4], Ramesh Hariharan[4], Minothi Parulekar[4], Karin M. Fredrikson[5], Timothy T. Harkins[5,6], Jeffrey Lin[2], Andrew B. Feldman[2], Pramila Tata[4], Georg B. Ehret[1], Aravinda Chakravarti[1]*

1 McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins School of Medicine, Baltimore, Maryland, United States of America, **2** Johns Hopkins University Applied Physics Laboratory, Laurel, Maryland, United States of America, **3** National Institute of Standards and Technology, Gaithersburg, Maryland, United States of America, **4** Strand Life Sciences, Bangalore, India, **5** Roche Diagnostics, Indianapolis, Indiana, United States of America, **6** Life Technologies, Beverly, Massachusetts, United States of America

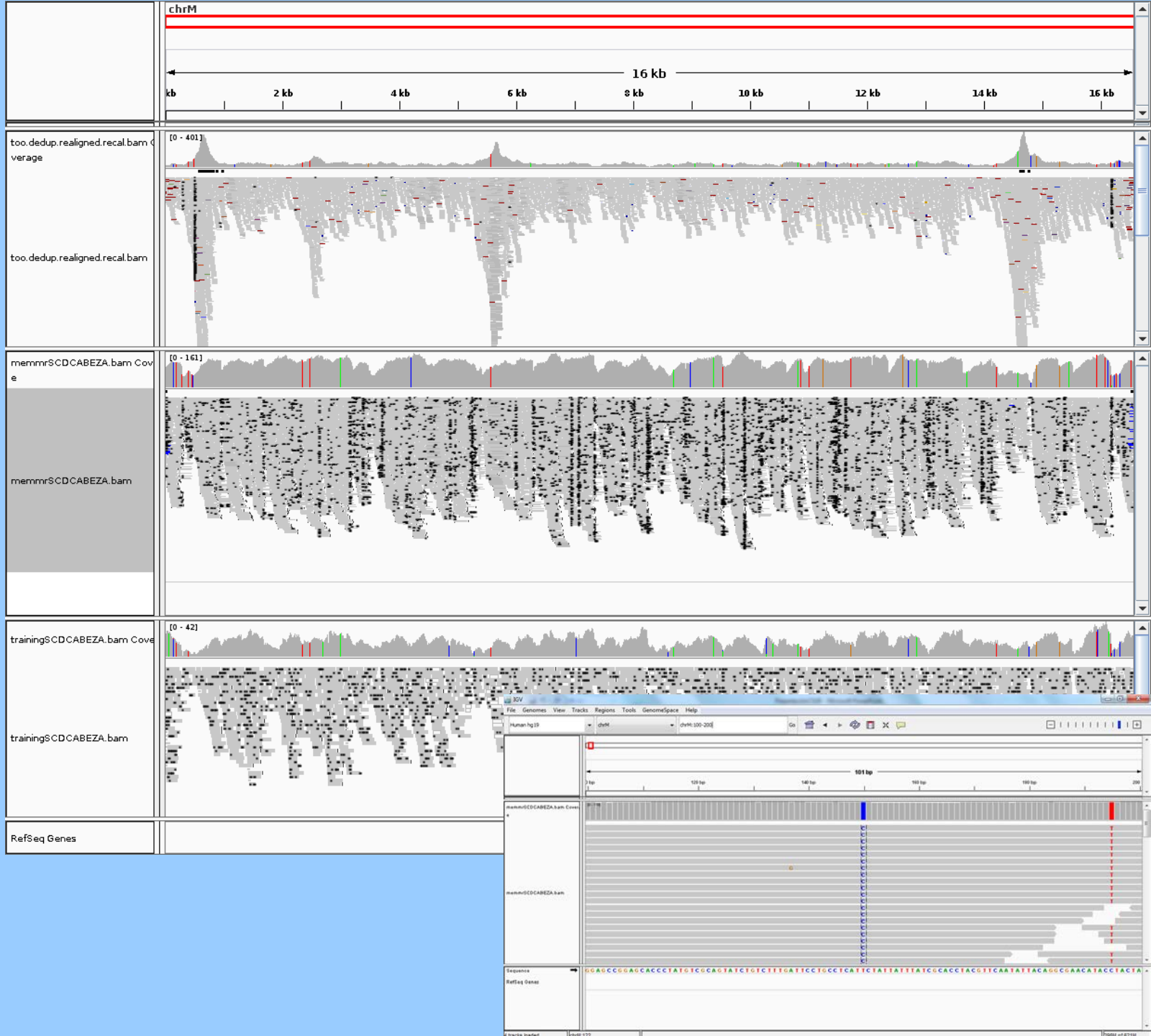
Abstract

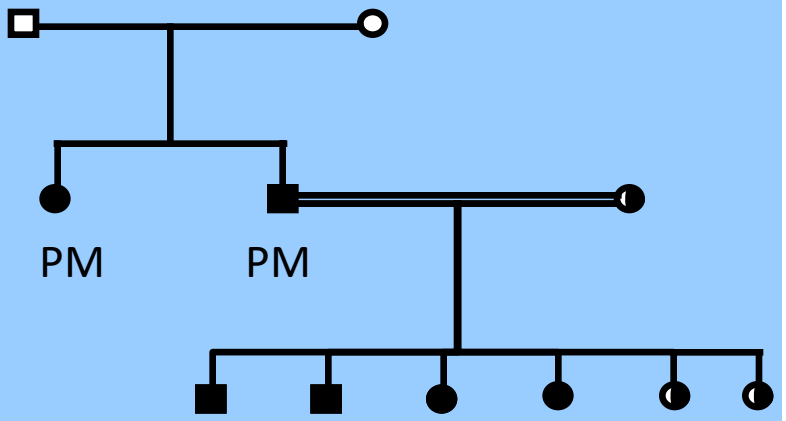
We describe methods for rapid sequencing of the entire human mitochondrial genome (mtgenome), which involve long-range PCR for specific amplification of the mtgenome, pyrosequencing, quantitative mapping of sequence reads to identify sequence variants and heteroplasmy, as well as *de novo* sequence assembly. These methods have been used to study 40 publicly available HapMap samples of European (CEU) and African (YRI) ancestry to demonstrate a sequencing error rate $<5.63\times10^{-4}$, nucleotide diversity of 1.6×10^{-3} for CEU and 3.7×10^{-3} for YRI, patterns of sequence variation consistent with earlier studies, but a higher rate of heteroplasmy varying between 10% and 50%. These results demonstrate that next-generation sequencing technologies allow interrogation of the mitochondrial genome in greater depth than previously possible which may be of value in biology and medicine.

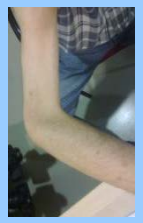
EXOMA

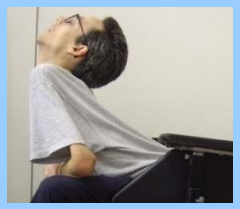
PLAQUETAS

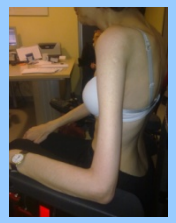
GENOMA

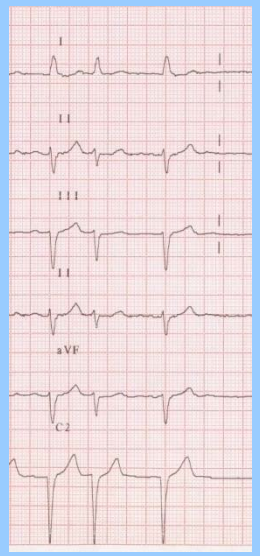


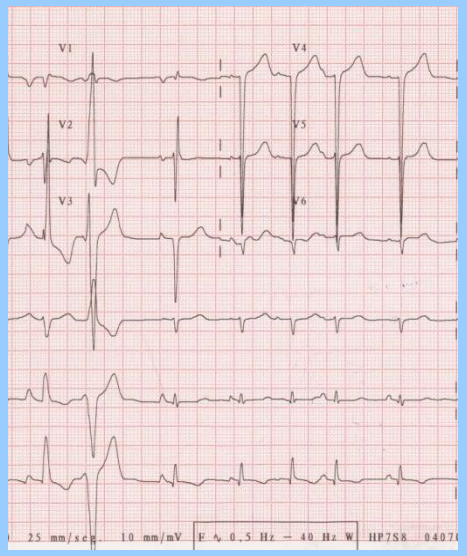












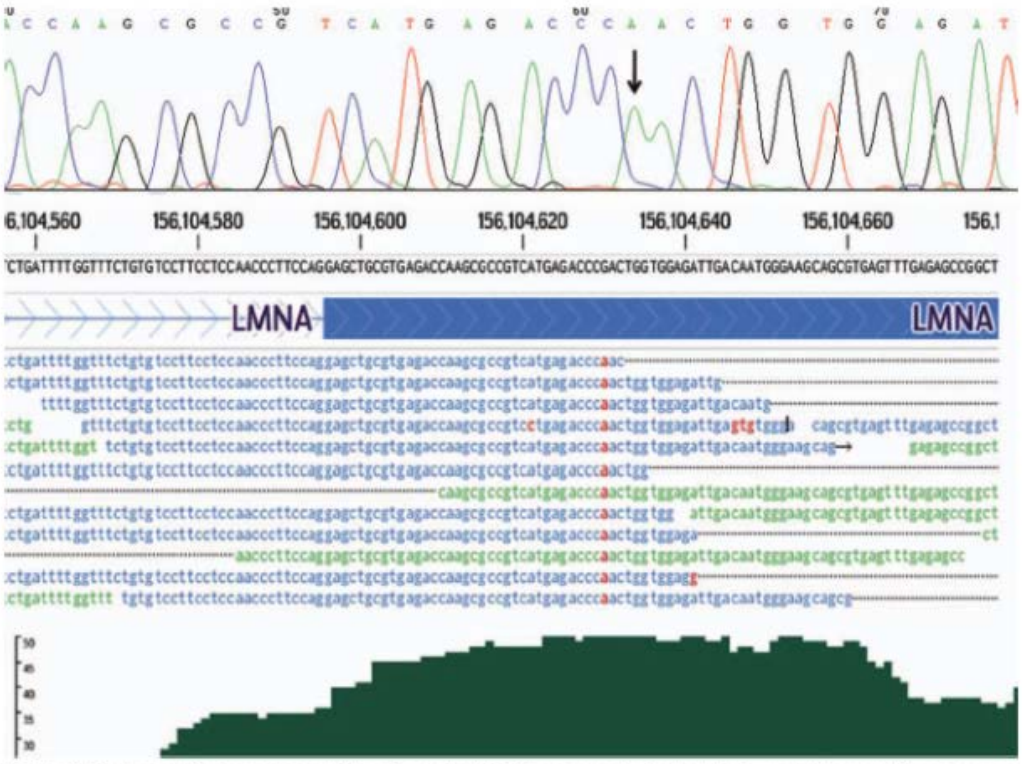


FIGURE 3. Next-generation sequencing multiple alignments at *LMNA* gene exon 4, showing the G→A mutation; and Sanger sequencing at this locus (top). [Color figure can be viewed in the online issue, which is available at wileyonlinelibrary.com.]

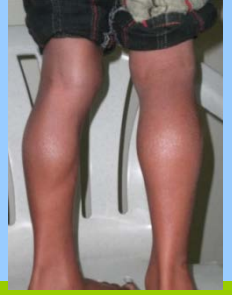
CASE OF THE MONTH

AUTOSOMAL RECESSIVE EMERY-DREIFUSS MUSCULAR DYSTROPHY CAUSED BY A NOVEL MUTATION (R225Q) IN THE LAMIN A/C GENE IDENTIFIED BY EXOME SEQUENCING

ADRIANO JIMENEZ-ESCRIG, MD, PhD,[1,2] ISABEL GOBERNADO, MD,[2,3] MERCEDES GARCIA-VILLANUEVA, MD, PhD,[4] and ANTONIO SANCHEZ-HERRANZ, BS, PhD[2,5]

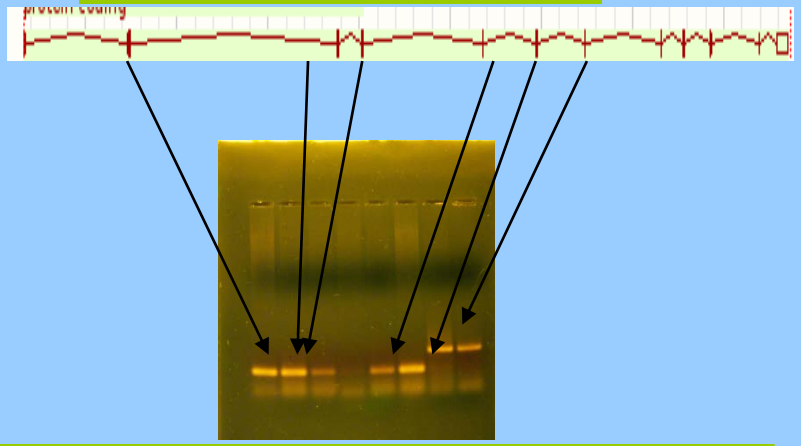
[1] Servicio de Neurologia, Hospital Ramon y Cajal and Universidad de Alcala, 28034 Madrid, Spain
[2] Unidad Central de Apoyo a Estudios Genomicos, IRYCIS, Madrid, Spain
[3] Servicio de Psiquiatria, Hospital Ramon y Cajal, Madrid, Spain
[4] Servicio de Anatomia Patologica, Hospital Ramon y Cajal, Madrid, Spain
[5] Servicio de Neurobiologia, Hospital Ramon y Cajal, Madrid, Spain

Accepted 10 October 2011



SECUENCIACION POR SANGER

Next Generation Sequencing (NGS)

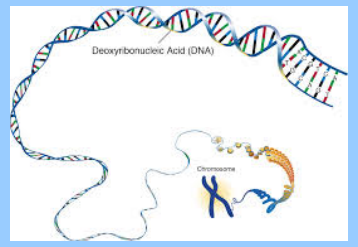


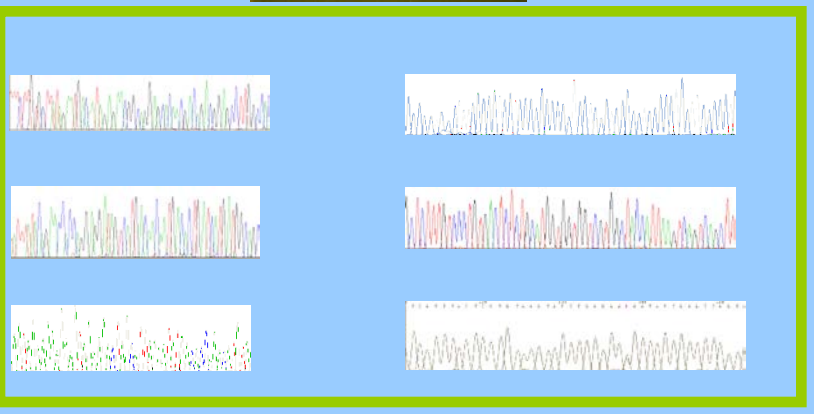
500-1000 €

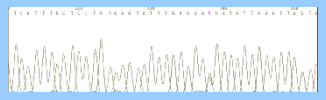
2 / 3 – 10 dias

1-2 / 27.000 genes

10-50 / 300.000 exones







genoma

Captura: panel de genes
exoma