Els documentalistes I la HC electròmica

By **JRCAT** may'15

•Els documentalistes gestionem HC

- Paper, electròniques,.....No històries personals.

• Nova informació en un nou format

- "What is genetic testing? - Genetics Home Reference". Ghr.nlm.nih.gov. 2011-05-30. Retrieved 2011-06-07.

- Genetic testing is "the analysis of chromosomes (DNA), proteins, and certain metabolites in order to detect heritable disease-related genotypes, mutations, phenotypes, or karyotypes for clinical purposes."[5] It can provide information about a person's genes and chromosomes throughout life

If you can't afford $1000 to know more about your genetic destiny, then store your DNA at home! DNA Direct makes it possible for you.

Image credit and more info: Eye on DNA

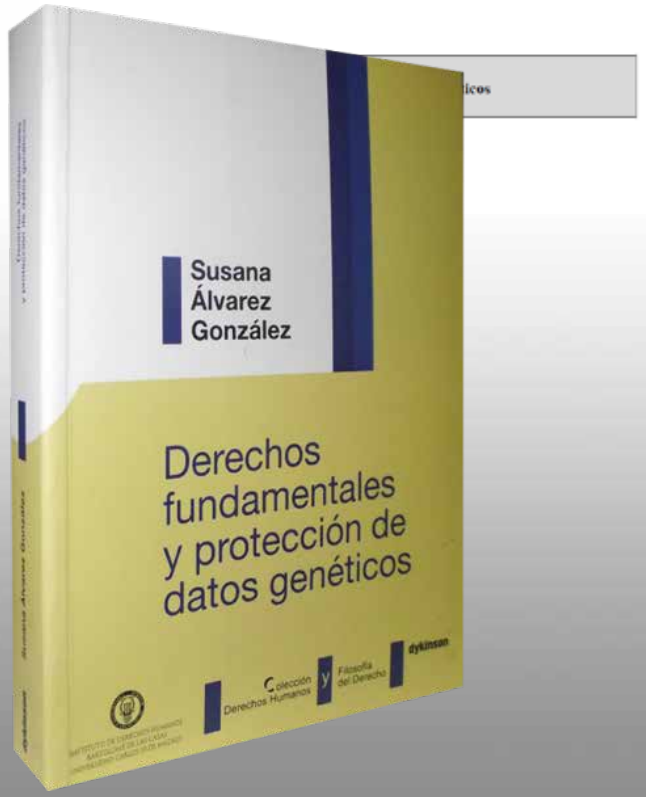
If yes, Medgadget's tip could be the best option:

• Especial protecció

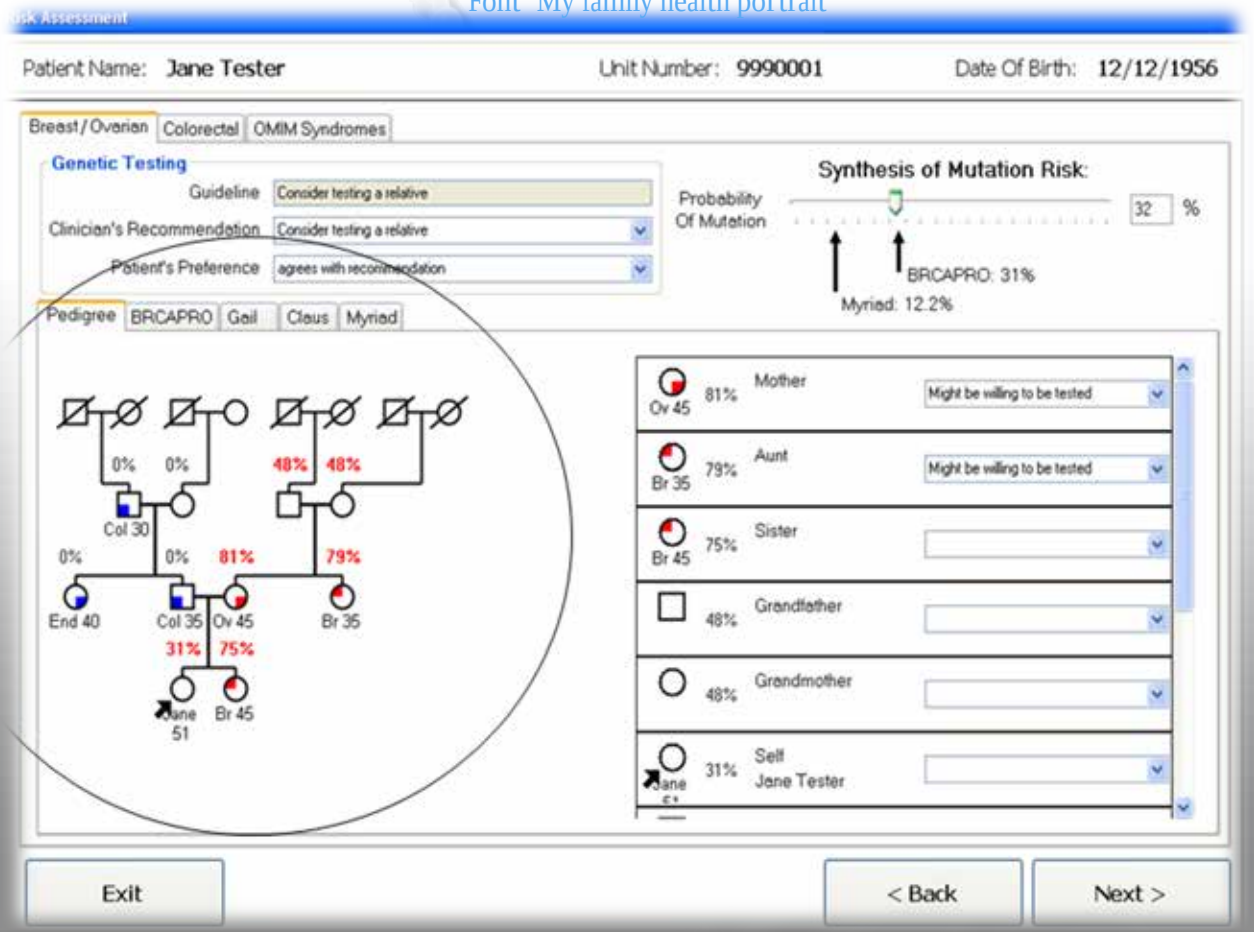
• Impacte a família

Grupo del artículo 29 sobre protección de datos

12178/03/ES
WP 91

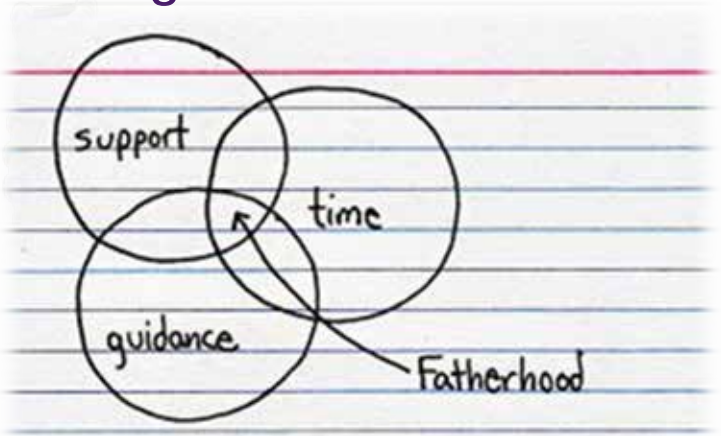


Font “My family health portrait”



• Compte amb la interpretació

- Quina educació teniu com a metges??? I ciutadans??



Then don't forget to browse your own SNPs gene by gene.

<<< Back to your Whole Genome

Jump to a gene: Go a SNP: Go

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

4

191M Bases
1157 Genes
34k SNPs

Your data includes **34525** SNPs on chromosome **4**.

Prev | 1 of 1381 pages | Next

	Gene	Position	SNP	Ver
⊞	LOC389833, ZNF595, ZNF718, LOC727739, LOC727764	63508	rs4690284	C o
⊞	LOC389833, ZNF718, LOC727764	80883	rs11724335	C o
⊞	LOC389833, ZNF718, LOC727764	84380	rs12509346	A o
⊞	ZNF718, LOC727764	92571	rs9884834	C o
⊞	ZNF718, LOC727764	100646	rs11727494	A o
⊞	ZNF718, LOC727764	111652	rs9996597	C o

Tell Me About...
...the Genome Explorer.
...SNPs without genes.
...NC vs. NG.
...rs numbers.
...SNP strand.

Indications

- Indication: Profound sensorineural hearing loss

Specimen and Genomic Source Class

- Peripheral Blood
- Genomic source class: Germline

Summary of Tests Performed

- GJB2 Full Gene Test
- GJB6-D13S1830 deletion
- Mitochondrial Hearing Loss Mutation Test

Overall Interpretation

- **Inconclusive.**
- DNA sequencing detected two changes in the GJB2 gene, 79G>A (V27I) and 109G>A (V37I). The V27I change has been reported as a benign variant (reference and is not believed to cause hearing loss. The V37I mutation has been previously reported in patients with hearing loss. This mutation, in homozygosity or combined with another GJB2 disease causing mutation, typically results in a mild to moderate hearing loss (Cryns et al. 2005). Mutations in both copies of the GJ gene are necessary to assume that GJB2 is responsible for the hearing loss. Although two mutations were identified in this patient, we would assume that the combination of a benign variant and a mild pathogenic mutation would result in a mild to moderate hearing loss rather than a moderately-severe one, as in this patient. It is most likely that the hearing loss in this patient is the result of the V37I mutation and an unknown second pathogenic mutation. It should be noted th second mutation is not identified in a large percentage (10-50%) of patients with nonsyndromic hearing loss and GJB2 mutations (del Castillo et al. 2003).
- GJB6-D13S1830 Deletion: A PCR-based analysis of the GJB6-D13S1830 region of chromosome 13 was performed and did not detect the deletion. This test does assess the DNA sequence of the GJB6 gene or detect other mutations that could affect the expression of the gene.
- Mitochondrial Hearing Loss mutations: Targeted bidirectional sequencing of mitochondrial DNA 1555 and 7445 regions did not detect the presence of these mutations.

Recommendations

- Although some cases may represent a coincidental carrier state, all of the studies have concluded that there are likely to be other genetic mutations that have n yet been identified. Genetic counseling is recommended for this patient and his/her family members.

At last, download your own data which would take about 5MB.

•Que estem fent?

- **Consentiment,** escanejar, donar en ma, guardar a part,..
- **Codificar??**

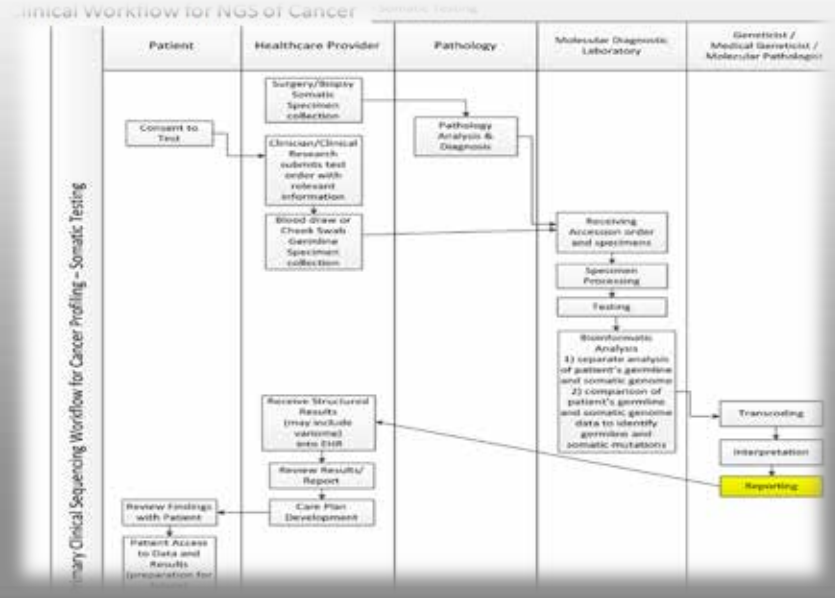
- Més de 2.000

☐Test diagnòstics per adequar tractament

☐Test de suport reproductiu

☐Tests predictius

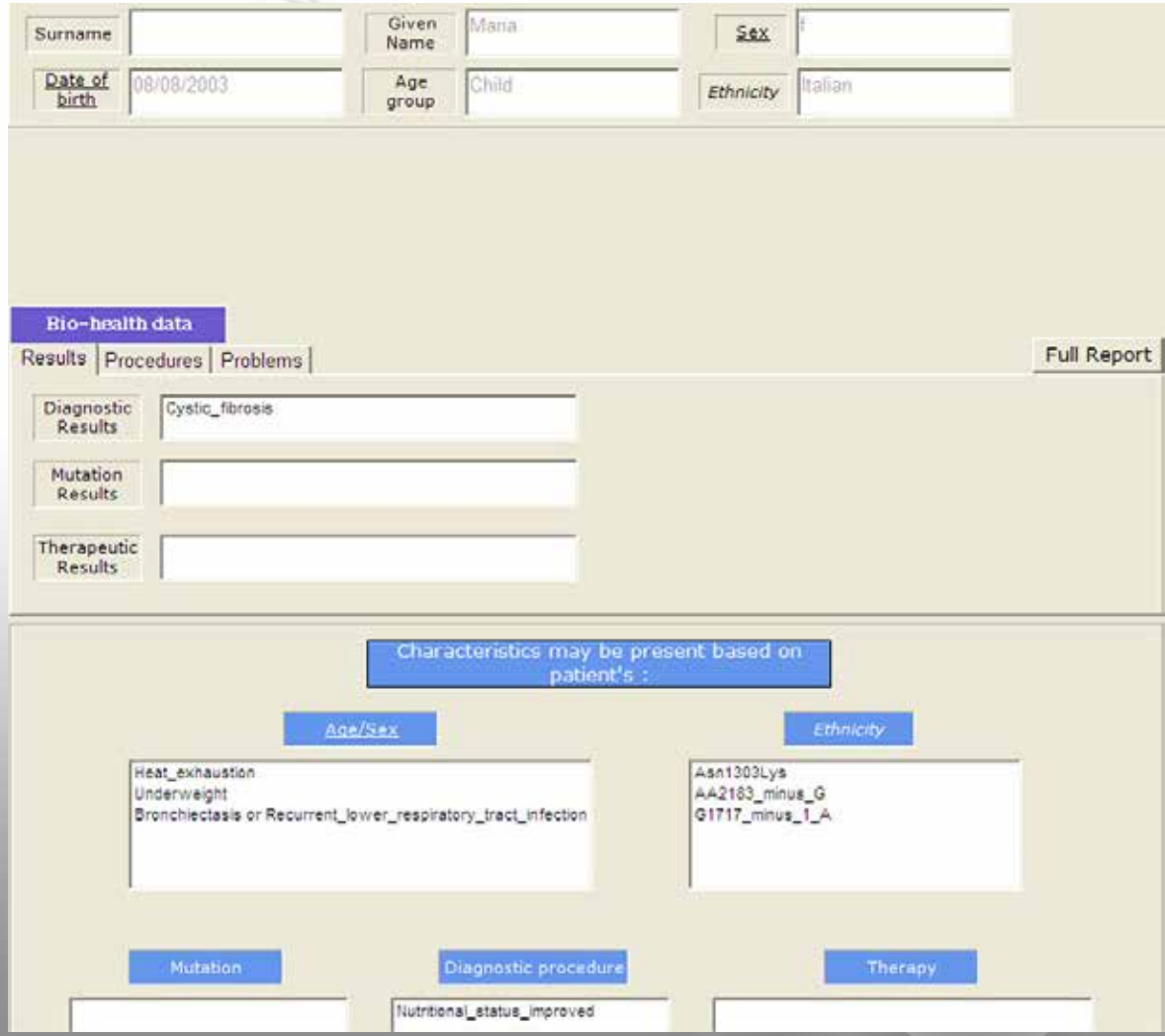
"Nomenclature of Medicine-Clinical Terms **(SNOMED-CT**) has a strong representation of ***clinical conditions***, The Logical Observation Identifiers Names and Codes **(LO-INC)** provides reasonable coverage of possible genetic ***test procedures*** and the Clinical Bioinformatics Ontology **(CBO)** provides a semantically structured vocabulary representing the ***physical observations*** of the diagnostic laboratory (variants observed, cytogenetic findings etc.)"



•Que tenim que fer

- A més de complir amb la legislació, i els PNT interns

- **UN PROJECTE DINS LA SCDM**
- **Entrevistes semiestructurades** a professionals. Tractarem temes de custodia i confidencialitat, organització de la info òmica i reptes i oportunitats a la hora d' integrar aquesta info a la hc3
- Pensem en uns **mix de participants:** documentalistes, metges assistencials, genetistes , bionformàtics i representants legals i de sistemes d' informació



Gràcies per la vostra atenció

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Veure:

HL7 Clinical Genomics Group. HL7 clinical genomics. <http://www.hl7.org/

Special/committees/clingenomics/index.cfm>

http://www.informaticsreview.

com/wiki/index.php/Confidentiality,_privacy,_and_security_of_genetic_and_ge

nomic_test_information_in_electronic_health_records:_points_to_consider

Sequeiros J, Paneque M, Guimaraes B. et al. The wide variation of definitions of

genetic testing in international recommendations, guidelines and reports. J Community

Genet. 2012;3(2):113–124. [PubMed]

Ronquillo JG, Li C, Lester WT. et al. Genetic testing behavior and reporting patterns in

electronic medical records for physicians trained in a primary care specialty or

subspecialty. Am Med Inform Assoc. 2012 Apr 17; [Epub ahead of print] [PubMed]