

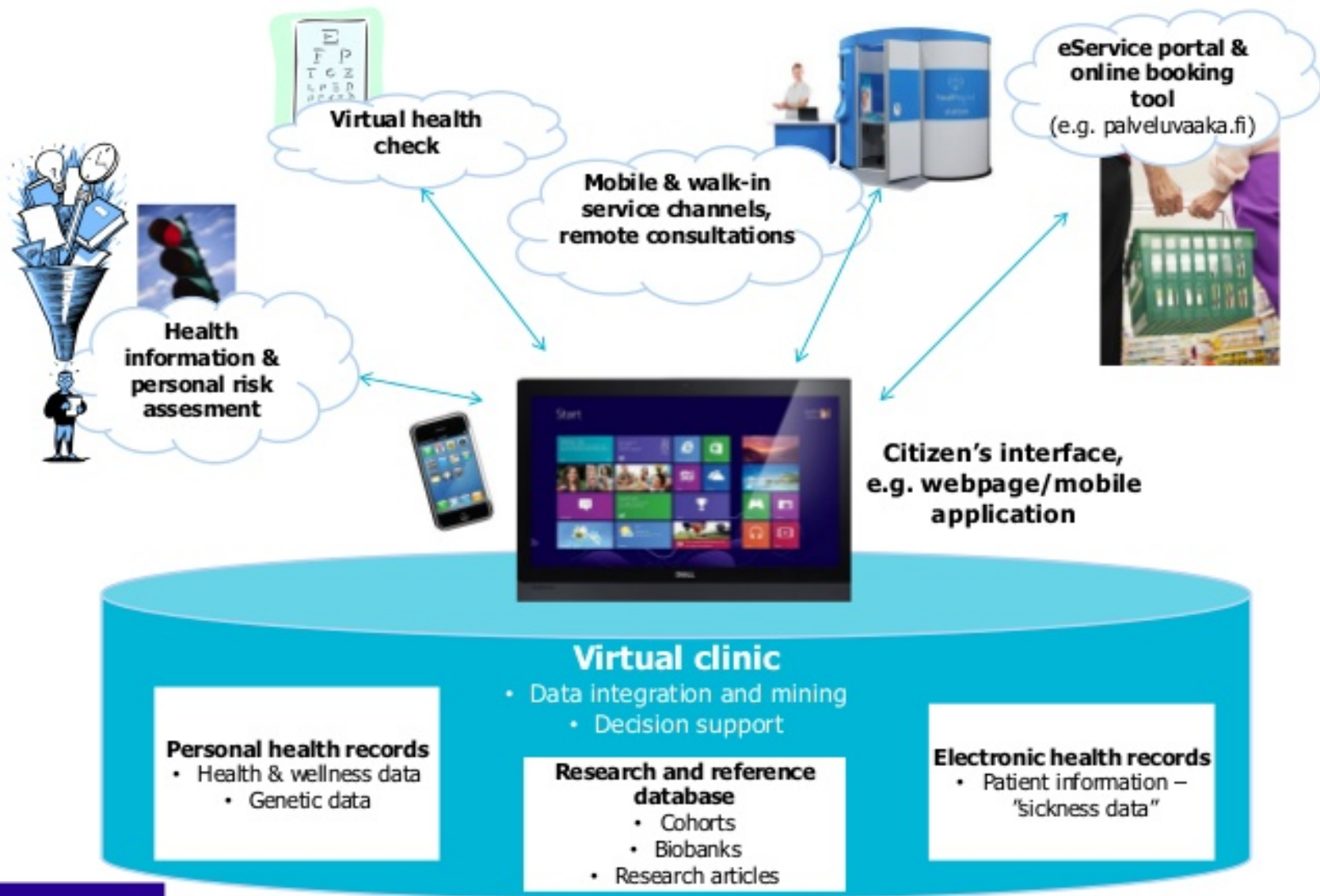


Els documentalistes i la HC electrònica

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"

Risk Assessment

Patient Name: Jane Tester Unit Number: 9990001 Date Of Birth: 12/12/1956

Breast/Ovarian Colorectal OMIM Syndromes

Genetic Testing

Guideline: Consider testing a relative
Clinician's Recommendation: Consider testing a relative
Patient's Preference: agrees with recommendation

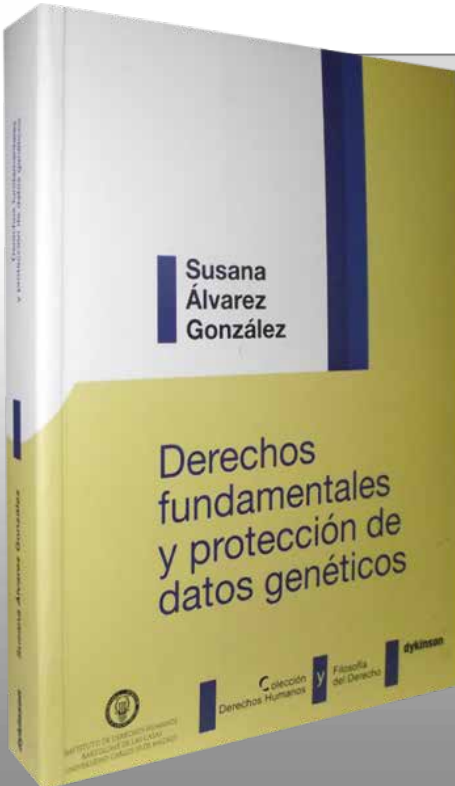
Probability Of Mutation: 32 %

Synthesis of Mutation Risk:
Myriad: 12.2%
BRCAPRO: 31%

Pedigree: BRCAPRO Gail Claus Myriad

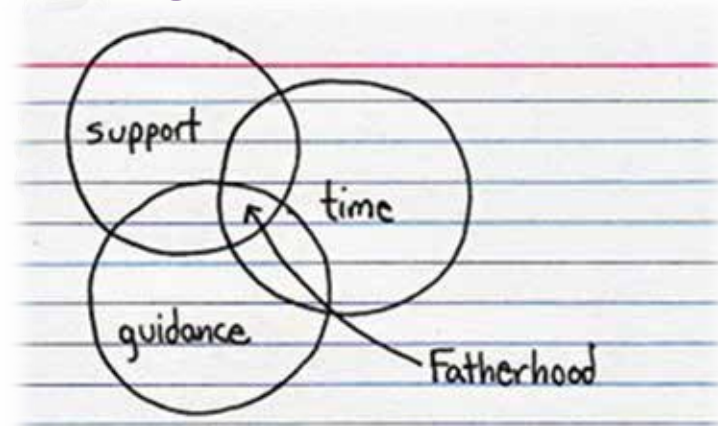
<input checked="" type="checkbox"/>	81%	Mother	Might be willing to be tested
<input checked="" type="checkbox"/>	79%	Aunt	Might be willing to be tested
<input checked="" type="checkbox"/>	75%	Sister	
<input type="checkbox"/>	48%	Grandfather	
<input type="checkbox"/>	48%	Grandmother	
<input checked="" type="checkbox"/>	31%	Self	Jane Tester

Exit < Back Next >



• 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, LOC727784	63508	rs4690284	C OI
LOC389833, ZNF718, LOC727784	80883	rs11724335	C OI
LOC389833, ZNF718, LOC727784	84380	rs12509346	A OI
ZNF718, LOC727784	92571	rs9884834	C OI
ZNF718, LOC727784	100646	rs11727494	A OI
ZNF718, LOC727784	111652	rs9996597	C OI

Tell Me About...

- ...the Genome Explorer.
- ...SNPs without genes
- ...NC vs. NG
- ...rs numbers
- ...SNP strand

Summary

Indications

- Indication: Profound sensorineural hearing loss

Specimen and Genomic Source Class

- Peripheral Blood
- Genomic source class: Gemline

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 GJB2 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 this second mutation is not identified in a large percentage (10-50%) of patients with nonsyndromic hearing loss and GJB2 mutations (de la Castilla 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 not 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 not 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

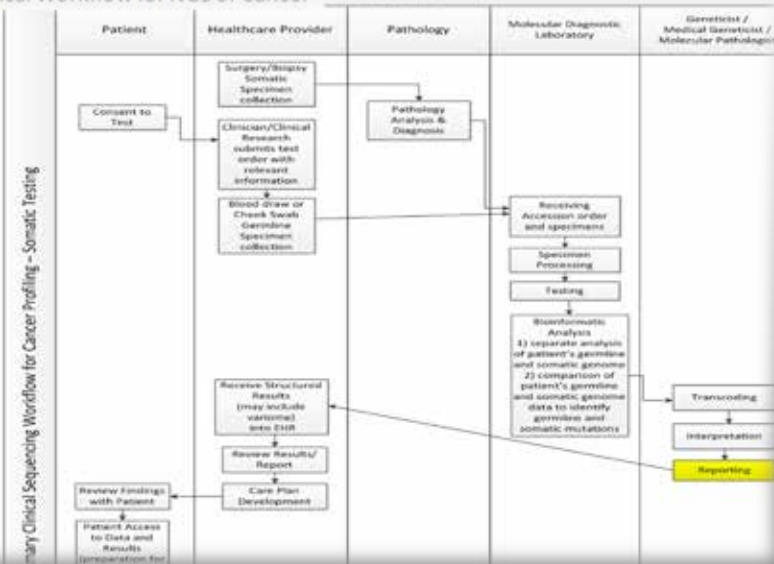
“Nomenclature of Medicine-Clinical Terms (SNOMED-CT) has a strong representation of *clinical conditions*, The Logical Observation Identifiers Names and Codes

(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.)”

Clinical Workflow for NGS of Cancer

Somatic Testing



The screenshot shows the OMIM website interface. The header includes the NCBI logo and the OMIM title. The search bar is active, and the search results page displays the OMIM title and a welcome message. The search results include a list of search terms and a list of search options (Limits, Preview/Index, History, Clipboard, Details). The search results also include a list of search terms and a list of search options (Limits, Preview/Index, History, Clipboard, Details).

• 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 custòdia 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, bioinformàtics i representants legals i de sistemes d' informació

The image shows a screenshot of a patient data form. At the top, there is a header with a small figure of a person. Below the header, there are several input fields for personal information: Surname, Given Name (Mana), Sex, Date of birth (08/08/2003), Age group (Child), and Ethnicity (Italian). Below this, there is a section titled "Bio-health data" with tabs for Results, Procedures, and Problems. Under the Results tab, there are three input fields: Diagnostic Results (Cystic_fibrosis), Mutation Results, and Therapeutic Results. At the bottom, there is a section titled "Characteristics may be present based on patient's :" with two columns: Age/Sex and Ethnicity. The Age/Sex column lists Heat_exhaustion, Underweight, and Bronchiectasis or Recurrent_lower_respiratory_tract_infection. The Ethnicity column lists Asn1303Lys, AA2183_minus_G, and G1717_minus_1_A. Below this, there are three tabs: Mutation, Diagnostic procedure, and Therapy. The Diagnostic procedure tab is active, showing Nutritional_status_improved.

Surname		Given Name	Mana	Sex	
Date of birth	08/08/2003	Age group	Child	Ethnicity	Italian

Bio-health data

Results | Procedures | Problems | [Full Report](#)

Diagnostic Results	Cystic_fibrosis
Mutation Results	
Therapeutic Results	

Characteristics may be present based on patient's :

Age/Sex	Ethnicity
Heat_exhaustion Underweight Bronchiectasis or Recurrent_lower_respiratory_tract_infection	Asn1303Lys AA2183_minus_G G1717_minus_1_A

Mutation | **Diagnostic procedure** | **Therapy**

	Nutritional_status_improved	
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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_genomic_test_information_in_electronic_health_records:_points_to_consider

[com/wiki/index.php/Confidentiality,_privacy,_and_security_of_genetic_and_genomic_test_information_in_electronic_health_records:_points_to_consider](http://www.informaticsreview.com/wiki/index.php/Confidentiality,_privacy,_and_security_of_genetic_and_genomic_test_information_in_electronic_health_records:_points_to_consider)

[com/wiki/index.php/Confidentiality,_privacy,_and_security_of_genetic_and_genomic_test_information_in_electronic_health_records:_points_to_consider](http://www.informaticsreview.com/wiki/index.php/Confidentiality,_privacy,_and_security_of_genetic_and_genomic_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]

