



Understanding the Needs of Consumers in the Use of Genome-based Health Information Services

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Clinical Validation (deCODE genetics)

- The genetic risk test for each disease is well-validated
- Each genetic marker has been replicated in 5 to 60 separate populations
- The relative risk is derived from several thousands of patients and tens of thousands of controls – much larger than the data used to support most FDA-approved diagnostics and drugs
- Markers are shown to be independent and are therefore combined by multiplying the risks together using a standard, validated method

Useful genetic risk tests for common disease – are we there yet?

- Heart attack – deCODE MI (2 markers)
 - 21% of the general population has 1.5 fold risk for early heart attack and 1.3 fold risk regardless of age of onset (comparable to risk conferred by LDL-cholesterol)
- Prostate cancer – deCODE ProCa (8 markers)
 - 10% of the general population has average 2 fold risk, 1% has 3 fold risk (lifetime risk 48%)
- Breast cancer – deCODE BreastCa (8 markers)
 - 5% of the general population has 2 fold risk, 1% 3 fold risk (lifetime risk of 36%)
- Type 2 Diabetes - deCODE T2 (4-8 markers)
 - 10% of prediabetic patients have two-fold risk for quickly progressing to type 2 diabetes (50 to 70% absolute risk within 3 to 4 years)
- Atrial fibrillation – deCODE AF (2 markers)
 - 25% of the general population has average two-fold risk of AF (1.5 to 3.6 fold)
 - Reveals that AF is a much greater cause for stroke than previously recognized
 - 150,000 stroke and TIAs per year may be due to undiagnosed AF - discharged on the wrong drug for secondary stroke prevention
 - Focusing extra cardiac monitoring on test-positive patients may save Medicare almost 1 Billion dollars after accounting for costs of tests
- Glaucoma – deCODE Glaucoma (3 markers)
 - 2.3 fold risk for more aggressive form of glaucoma leading to earlier blindness if unrecognized

A Case Study Demonstrating Usefulness of a Genetic Risk Test

- 48 year old white male in apparent good health,
 - His father diagnosed with localized prostate cancer at age 70
 - Patient with low normal PSA at age 42
- deCODE Prostate Cancer test results (8 markers combined)
 - Relative risk = 1.88 fold compared to general population risk for white males.
 - Calculated lifetime risk = $1.88 \times 16\% = 30\%$
 - Modestly higher risk for aggressive vs non-aggressive disease
- High risk prompted early serum PSA test by primary care
 - PSA was high normal at 2.5ng/ml
- High risk prompted referral to urologist
- High risk prompted urologist to perform ultrasound-guided biopsy
 - Positive for cancer in 3 out of 12 core biopsies – 15% volume
 - Intermediate grade tumor (Gleason score of 6)
- Surgery after negative workup for metastasis – likely cure