GENETICS, PHILOSOPHY

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I am currently writing a piece on genetic testing, basically arguing that genetic testing is still a research method and whole genome sequencing nothing for prime time as basically now summarized also in JAMA:

> In this exploratory study of 12 volunteer adults, the use of WGS was associated with incomplete coverage of inherited disease genes, low reproducibility of detection of genetic variation with the highest potential clinical effects, and uncertainty about clinically reportable findings. In certain cases, WGS will identify clinically actionable genetic variants warranting early medical intervention. These issues should be considered when determining the role of WGS in clinical medicine.

Maybe the judgment of any scientific method was largely limited to experts about 20 years ago. You had to know something about research, you had to go to a library, you had to find the relevant information and eventually put it into the right context. Only a few people and only a few journalists could do that. (and only the latter would even publish their opinion).
This has completely changed with so many research papers now being published online. There is no more gate, no more gatekeeper. It means, however, that research papers are frequently misinterpreted – from patient advocacy groups to companies to medical doctors. I would wish that research papers would carry a “For research use only!” label as printed on many bottles with enzymes, antibodies and alike (Medical information is otherwise still restricted in Germany to physicians, pharmacies and medical staff). Given that rather muddle-headed situation in genetic testing, I think the new JAMA paper is a welcome recommendation for everybody!

> incomplete … low reproducibility .. uncertainty

yea, yea.