

reported in *Science Translational Medicine* last week that they had used sequencing in newborns to sift for rare genetic mutations that might cause disease (C. J. Saunders *et al. Sci. Transl. Med.* 4, 154ra135; 2012). The results were impressive. For three of the four infants, probable culprits were identified.

To reach these conclusions, the team considered not just genetic regions in the babies, but also those in their relatives and in the scientific literature. However, for sequencing to reach its medical potential, researchers must be able to access even more genomes. Each person has millions of genetic variants — or sequences that differ from the human reference genome — making it hard to find those that might affect health. The key is to locate variants that recur in people with similar illnesses.

With so many sequencing projects under way, clinicians are always eager to know whether a variant has been observed in patients at other institutions. Analysis tools are available to help (see page 157). Yet there is currently no quick, reliable or convenient way to spread this information.

Data sharing through scientific publication has fuelled an impressive collection of databases that reveal frequencies of common variants. When variants or genes have been associated with disease, those results are also deposited in databases. ClinVar, a database from the US National Institutes of Health (NIH), for instance, gathers health-related genetic variations from the literature. And the NIH has set money aside to create a separate resource for clinically relevant genetic variants: essentially a curated database of variants for which some sort of clinical action is advised.

These are valuable efforts, but are inherently limited. Publication is too slow, and data collected about many variants will never be published. Researchers need to be able to query not just variants in the literature, but also those that have been found in other patients but not reported.

This January, an advisory group to the UK Department of Health said that the country should create a centralized facility to store genomic data

to improve treatments and diagnoses. However, in the United States, where many sequencing projects are based, regulations about sharing patient data will make setting up a centralized repository more difficult.

One option would be to give patients their own sequenced genome data, letting them deposit it where they choose. Already, 23andMe, a consumer-genetics company in Mountain View, California, has used data and DNA supplied by its customers to discover (and, controversially, to patent) disease-associated variants.

Another option is for medical-research institutions to agree on ways to share information with each other. Rather than transferring a full medical record, for example, a researcher at one institute could learn whether a variant had been observed in other people and, if so, what diseases they had. For this to work, new technology and shared platforms would have to be developed.

There are other problems. Sequencing data are imperfect. High-throughput sequencing technology sometimes overlooks variants or makes other errors. Downstream issues are rife: are the benefits of sequencing worth the costs? Can the information be protected? How accurate are conclusions? Should information not related to the immediate medical question be shared with patients, even if a diagnosis is uncertain or no treatment is available?

All of these questions would be easier to answer if genomics data could provide more certainty. Yet to achieve this, researchers must look at the genomes and health information of more people. At this stage, the best path forward remains unclear. But for genomics to advance, the community must communicate. Institutions must consider not just what is best for their particular situation, but also what is best for the broader community. A good place to start is for staff at genomics centres and hospitals to meet in person, to share experiences and best practices. ■

“There is currently no quick, reliable or convenient way to spread genomic information.”

Fighting chance

Collaboration between geneticists and economists has the potential to bear fruit.

One side is accused of supporting ethnic cleansing; the other of being intellectually naive. Does that sound like the beginning of a fruitful collaboration? Perhaps not, but read on.

As we report on page 154, an increasingly bitter spat has emerged between geneticists and economists over a paper that links a country's genetic diversity to its economic development.

At its heart, the argument boils down to cold statistics and methodological differences. A team of prominent geneticists and anthropologists at Harvard University in Cambridge, Massachusetts, says that the paper's economist authors did not properly account for historical and cultural connections between genetically similar countries, so correlations are mistaken for cause.

The work is part of an emerging trend to blend economics with genetics. Daniel Benjamin, an economist at Cornell University in Ithaca, New York, who is trying to identify the genetic basis for economically relevant traits such as risk aversion, is among those who say that the combination has yet to prove its worth. Nonetheless, he and others assert that understanding how genetics influences individual and international economies has the potential to inform policy.

For this to happen, both sides must take seriously the standards, methodology and history of the other. Geneticists have spent years grappling with the difficulties of getting useful information out of genomes. They have made mistakes, and learned from them, and it

is naive for social scientists to think that they are immune from these errors, or that they can learn all they need to quickly. Benjamin says that nearly every study that links individual economic traits with specific genetic variants, for example, is riddled with false positives.

Social scientists should also remember that human geneticists bear the historical scars of eugenics, and more recent accusations of insensitivity to indigenous populations. Any whiff of biological determinism will draw a strong response.

Geneticists, for their part, should acknowledge that quantitative social scientists are experts in measuring human behaviour, both individual and collective. An entire subfield of economics, called econometrics, exists to make sense of data that are just as seemingly random as the string of As, Ts, Cs and Gs that comprises a genome. Moreover, many of the statistical methods that economists now use have their roots in the work of early-twentieth-century geneticists. Closer collaboration between the two fields could unlock the knowledge and expertise of social scientists, enabling them to draw conclusions that geneticists would never have conceived.

One hopeful model is the Social Science Genetics Association Consortium, a collaboration between social scientists, geneticists and epidemiologists that aims to bring more rigour to the search for the genetic basis of economic and other behavioural traits. In addition to combining the expertise of scientists in disparate fields, the consortium also has access to dozens of cohorts, encompassing more than 100,000 people.

The consortium was formed after Benjamin's team uncovered a genetic variant linked to educational attainment in some 2,000 Icelanders, only to find that the association could not be replicated in other populations, raising questions over whether it is real. The group's expertise and infrastructure give it a chance of finding genuine links that will hopefully see geneticists working on follow-up studies, rather than writing angry letters. ■

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