GENOMICS NETWORK

THE NEWSLETTER OF THE ESRC GENOMICS NETWORK

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ESRC genomics policy and research forum

The **Genomics Policy and Research Forum** is based at Edinburgh University

Edited by Ginny Russell (Egenis), Julie Hamilton (Innogen), Anne Wilbourn (Cesagen)

Design randhdesign
www.randh.net

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Welcome to the first Genomics Network newsletter

Over the last two years, the Economic and Social Research Council has funded five new initiatives throughout the UK, providing Britain's social scientists with a potentially unrivalled reputation in genomics research. From GM crops to genetic testing, every possible application of genomics raises new and exciting questions. Recent leaps in the scientific study of genes and our growing ability to manipulate the genomes of plants, animals and humans far outstrips our understanding of the social and economic consequences of genomics.

The objective of the Genomics Network as a whole will be to undertake a systematic, critical and technically informed exploration of the past, present and future economic and social trajectory of genomics. It comprises three sister centres throughout the UK:

Innogen, the ESRC Centre for Social and Economic Research on Innovation in Genomics; **Cesagen**, the ESRC Centre for Economic and Social Aspects of Genomics and **Egenis**, the ESRC Centre for Genomics in Society. In addition, the ESRC's **Genomics Survey** aims to provide a basic understanding of the attitudes of the general public towards human, animal, plant and environmental genomics. Finally, the **Genomics Policy and Research Forum** was established in 2004 and will be responsible for assisting the research centres in the dissemination of research.

In this first newsletter you will find introductions from each member of the Network and details of what we have achieved so far. Subsequent newsletters will expand on our research in more detail. We shall address topical issues in the field and let you know about all our up and coming events.

INTRODUCING THE NETWORK

MICHAEL BANNER

Genomics Policy and Research Forum

The Genomics Policy and Research Forum is based in Edinburgh and is directed by Michael Banner. The Forum aims to exploit synergies between the existing centres and programmes in the Network and to build up partnerships between them. The Forum will encourage interaction with genomic scientists and assist the Network in its engagement with policy makers and the public.

JOYCE TAIT

Innogen *www.innogen.ac.uk*

The Innogen Centre brings together social scientists, technology and policy analysts, economists, lawyers and ethicists to study the far-reaching social and economic implications of advances in the life sciences. The Centre is a partnership between the University of Edinburgh and The Open University, and is directed by Joyce Tait. Now in its second year, the Innogen Centre will continue bridging the boundaries between natural and social sciences in a truly interdisciplinary manner.

RUTH CHADWICK

Cesagen *www.cesagen.lancs.ac.uk*

Cesagen is a Lancaster-Cardiff collaboration and is directed by Ruth Chadwick in Lancaster with Paul Atkinson as Associate Director in Cardiff. Cesagen is a multidisciplinary centre in which staff from social sciences and humanities work closely with natural and medical sciences to address the social, economic and policy aspects of developments in genomics. In the light of considerable national and international attention to these issues, and increased public debate, Cesagen aims to undertake a programme of public engagement as well as feeding its research into policy circles.

JOHN DUPRÉ

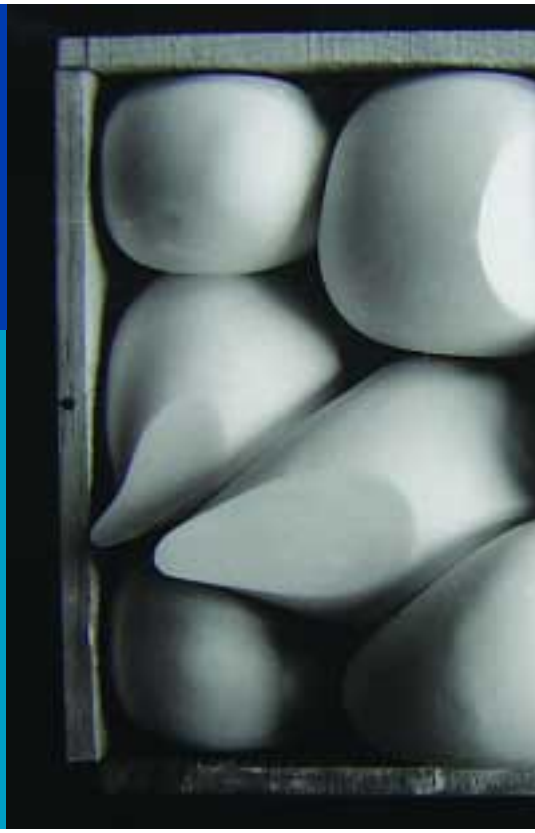
Egenis *www.ex.ac.uk/egenis*

Egenis is based at the University of Exeter and is directed by John Dupré. At Egenis we have a more theoretical approach to our research agenda than our sister centres; we are especially interested in the fundamental shift within the study of biology from thinking in terms of traditional genetics to working and thinking in terms of genomics. A central strand of our research addresses semantic issues arising from genomics; looking at the meanings of words that have become commonplace within the subject matter, as well as the development of the science itself. This conceptual work provides a strong and distinctive grounding for our social sciences research. Numerous articles and broadcasts about our work have appeared in the media and we have welcomed several new researchers.

Genomics Survey *www.surrey.ac.uk/SHS/genomics*

The Genomics Survey consists of a cross-disciplinary research team of sociologists and psychologists based in the School of Human Sciences at the University of Surrey. It is directed by Professor Richard Shepherd. The main aim of the Genomics Survey is to provide a basic understanding of current attitudes of the general public towards human, animal, plant and environmental genomics and how these attitudes are changed and formed over time.

What is genomics?



First, what is a genome? The genome of an organism is the entire genetic code of that organism. Most people know a little about DNA, genes, chromosomes, etc. - that they constitute the 'genetic information' that makes us what we are at birth. An organism's genome is all this stuff considered together.

Genomics is the science of genomes - more specifically, their sequencing, mapping, analysis, study and manipulation.
The reason for the new term is that the ability to consider the entire genetic information of an organism is relatively new, and opens up entirely new possibilities that go well beyond what has been understood by previous terms.

NEWS

EGENIS NEWS

Artist in residence

Deborah Robinson is the Egenis artist in residence who is attempting to translate our work on genomics into a visual piece of work. Deborah has published an on-line diary detailing her journey towards the work. This diary is intended to capture the dynamics of the exchange of ideas that takes place during the residency. She writes 'I want to make a visual record of the laboratory, the space within which information about the genome is generated (as a space of origin). The technology and also the specific light conditions in the laboratory play some part in the mediation of the social perception of the genome.' Her work will culminate in a major exhibition.

INNOGEN NEWS

Generation Scotland

The University of Edinburgh funded Sarah Parry, Sarah Cunningham-Burley, Jane Ewins, Ann Bruce and Graeme Laurie to carry out preliminary research on social, legal and ethical issues relevant to the Generation Scotland genetic database. This is being developed by Professor David Porteous, head of the Molecular Medicine Centre at Edinburgh University. Generation Scotland will address Scotland's three health priority areas - cancer, heart disease/stroke, and mental health. It should also shed light on the role of heritable (genetic) risk factors in early diagnosis, disease monitoring, treatment optimisation, avoidance of adverse drug reactions, healthcare planning and drug discovery. Another major project has also now been funded by SHEFC. This is entitled 'Genetic Health in the 21st Century' (21CGH) and will contribute to both Generation Scotland and Biobank. 21CGH is also funding two new fellowships for three years, one to be based in Innogen and one in the associated AHRB Research Centre in IP and IT Law.

CESAGEN NEWS

'Research Hotel' established at CESAGen

Cesagen has welcomed its first visitors to its 'Research Hotel'. This facility was set up to provide a physical location for visiting academics to come and work closely with Cesagen researchers for a short period of time. Cardiff have already welcomed Jane Karwoski from University of Nevada and Margaret Sleebom from The International Institute for Asian Studies, The Netherlands. Lancaster are currently playing host to Dr Georg Bosshard from Zurich University's Institute of Legal Medicine and Bert Gordijn, Nijmegen University, The Netherlands.

CONFERENCE REPORTS

Precaution and Progress: Lessons from the GM dialogue

The first Innogen International conference was held on 13 November 2003 in Edinburgh. Co-sponsored by the AEBC and the Scottish Executive, the conference was attended by 134 delegates and was judged a great success. A strong theme in discussions was the political advantage gained by those who had managed to categorise a wide range of developments under the single heading, GM crops. GM is a complex area and decisions about it raise fundamental issues facing nations and global societies. Future decision-making should consider all the arguments and involve engagement with a wide range of potential stakeholders.
The report of the conference is available from Innogen on request.

Genomics and Society

CESAGen held its first International Conference in March 2004 at The Royal Society in London. Attracting over 200 UK and international academics and researchers, the conference was an overwhelming success. The theme of 'Genomics and Society' addressed the main thrusts of the Centre's research agenda: the implications of genomics in the

below: Work by Dominic Hopkinson

transformations of knowledge production; economics and innovation; ethics and regulation; identity and social organisation; risk and responsibility; global discourse and cultural capital. In the plenary and general sessions presentations were given on the impacts of genomics developments in the areas of medicine, health, food and agriculture.

The conference abstract book is available on the website and we will also be publishing a book of some of the key papers.

Meanings of Genomics

Sir John Sulston launched the first Egenis conference in November 2003 with a brilliant keynote address looking at the transition from genetics to genomics, the sequencing of the human genome, and the subject of open access to information. The talk is available to all on our website. Over three days Egenis was pleased to welcome delegates and speakers from all over the UK, Europe and the USA. Workshops on 'what is a gene' and 'what is a genome' proved the perfect launch pad for the debates that followed.

The papers generated by the conference will form an entire issue of the Journal 'New Genetics and Society' early in 2005.

INNOGEN www.innogen.ac.uk

Innovation processes in genomics industry sectors
Research Fellow: James Mittra

Genomic and biotechnology partnerships in less developed countries
Research Fellows: Aparna Joshi and James Smith

Genomics innovation in Scotland
Research Fellow: Alessandro Rosiello

Institutional impacts of North-South partnerships in agricultural biotechnology
Research Fellow: Seife Ayele

The national and international policy environment for genomics
Research Fellow: Catherine Lyall

Interests and values in risk-related stakeholder interactions
Research Fellow: Ann Bruce

Genetic health in the 21st century (21CGH)
Research Fellow: Gill Haddow

Generation Scotland
Associated staff: Sarah Cunningham Burley, Sarah Parry, Gill Haddow

The social dynamics of public engagement in stem cell research
Associated staff: Sarah Parry

Farmers' understanding of genetically modified crops within local communities
Associated staff: Andy Lane, Susan Carr, Sue Orefzcyn

CESAGEN www.cesagen.lancs.ac.uk

Transcending the genome: the paradigm shift to proteomics
Senior Research Associate: Ruth McNally

Plant genomics, commercialisation and environmental knowledge
Research Associate: Katrina Stengel

Genetic databases
Research Associate: Minakshi Bhardwaj

The economics of innovation in genomics
Research Associate: David Knight

Genetics, health and identity
Research Associate: Susan Hogben

The emerging politics of new genetic technologies
Research Associate: Alex Plows

Indigenous peoples and globalisation of genomics in Amazonia
Research Associate: Paul Oldham

The media, culture and genomics
Research Associates: Joan Haran (Cardiff) Kate O'Riordan (Lancaster)

Reconfigurations of human-animal relations in genomics and beyond
Research Associate: Richard Twine

EGENIS www.exeter.ac.uk/egenis

Patenting and the utility of genomic knowledge
Research Fellow : Jane Calvert

Semantic drift in the dissemination of genomic knowledge
Research Fellow : Christine Hauskeller

Genomic technologies in the developing world
Research Fellow: David Reece

Genetic testing for common complex conditions
Research Fellow: Paula Saukko

Genetic models of group membership, intergroup relations and individual identity
Lead Researcher: Thomas Morton

Farm animals and genetic modification
Lead Researcher: Carol Morris

Philosophy of biology
Research Fellow: Stefan Müller

The integrity of living beings as a normative concept in bioethics
Research Fellow: Michael Hauskeller

Pharmacogenetics

drugs and treatments specifically tailored for our needs

Elisa Pieri & Sarah Wilson

cesagen

Some of us may think that there is nothing new in the idea of receiving drugs and treatments specifically tailored for our requirements. Increasingly however, big players in healthcare and drugs companies have gone on record admitting that sometimes 'our drugs don't work'.

The same pronouncements often include references to pharmacogenetics as the solution to that problem. Pharmacogenetics is the study of the genetically-determined variability in response to drugs, which is to say the study of how an analysis of our genes may help us in choosing the most appropriate drug, selecting the optimal dose, and hopefully avoiding the risk of adverse drug reactions.

The term may sound obscure, but with the Government recently allocating £4M for studies in this field, it is certainly here to stay. The 2003 Government White Paper entitled *Our Inheritance, Our Future* proclaims that 'pharmacogenetics will lead to prescribing which is more effectively tailored to the needs of the individual'. But will pharmacogenetics really deliver? And are there issues that we ought to be thinking about before or while we go down that path?

First and foremost is the question of whether the claims to personalised medicines are overstated, given that the evidence base is still relatively small, and an increasing number of conditions are thought to be the result of interactions of different (and often largely environmental) factors. On the other hand, adverse reactions and lack of response may also be caused by factors beyond our genetic make-up, such as interactions with other drugs or factors like age, liver and kidney functions, medical errors and patient non-compliance with a given treatment. Relevant questions in this context could be raised about the level of investment in developing this technology – is it justified, and how does it impact on the possibility of exploring alternatives? In other words, since it is ultimately funded by taxpayers' money, do we think it is a sensible allocation of resources?

Other complex issues are likely to be raised by the fact that pharmacogenetics presupposes individual genetic testing as the norm.

Concerns may be expressed about the clinical reliability and validity of testing, as well as questions of equity and fairness.

The quality of the testing would need to be checked along with its clinical performance and its relevance to the specific patient group that it is directed to. Drug testing still focuses mainly on white and Japanese people, as they make up the big markets for drugs, with the result that people in or from other regions continue to be disadvantaged. Pharmacogenetics has the potential to either exacerbate exclusion of certain ethnic groups from research and treatment, or conversely it could potentially secure a more appropriate predictor of response to treatment than traditional racial indicators based on appearances.

Pharmacogenetics could raise other ethical issues linked to access and equality by segmenting further the markets and producing a myriad of groups of patients or conditions that may not be sufficiently numerous to justify the costs associated with developing appropriate treatments (known as orphan drugs). Even when patients are thought to respond to a treatment, difficulties emerge when trying to decide thresholds indicative of a patient responding 'well enough', so as to justify and guarantee inclusion in the provision of that treatment. Furthermore, if we accept that there currently are inequalities in the provision of health services – geographical or otherwise – it is legitimate to be concerned that testing may also end up being unequally provided, posing increased

risks to those patients that might possibly be prescribed a pharmacogenetic treatment without the appropriate testing beforehand.

The greatest ethical concerns posed by pharmacogenetics though, are likely to be those linked to privacy and safeguards of confidentiality, consent to be tested, identification through genetic information, and storage of the sample provided for testing.

It is perhaps early days to be able to fully understand the potential repercussions and ramifications of those concerns. There are nonetheless a few areas in which debate has already started, particularly with reference to insurability, use of genetic information in employment policy, and implications of the potential for commercial exploitation.

The issue of how the results of genetic testing – which is a pre-requisite of pharmacogenetics - may be used by private insurers to exclude benefits from policies, to increase premiums (rendering policies unaffordable to many) or simply to refuse to insure people, also features prominently in the Government White Paper mentioned above. Similarly, the danger that individuals could in the future be denied employment opportunities – whether at recruitment, retention or promotion level – due to disclosure of their genetic information has also been recognised by the Government as real.

Debate also focuses on likely conflicts of interests arising from the commercial environment in which drugs are developed. It is likely that the same companies that manufacture pharmaceuticals will also be producing the tests that show whether those drugs are suitable for us or not.

In the midst of the uncertainty that surrounds pharmacogenetics, one thing *is* certain - there are a number of very pressing social, political and ethical issues that need to be addressed very urgently before this new technology can enter our daily lives and its applications find public acceptance.

Elisa Pieri and Sarah Wilson are research associates at the Northwest Genetics Knowledge Park (Nowgen) and Cesagen. They can be contacted at ***e.pieri@lancaster.ac.uk*** *and* ***s.e.wilson@lancaster.ac.uk***

Do genes exist?

egenis

Karola Stotz

The common perception of genes as special kinds of DNA sequences found on chromosomes between much longer stretches of 'junk DNA' is severely outdated. Sequences that play the traditional role of a gene, namely coding for a protein, make up less than 2% of human DNA, but the number of sequences that perform other functions is expanding rapidly. These functions range from more 'gene like' functions such as manufacturing one of the newly discovered classes of RNA which help to regulate genome expression, to very un-gene-like tasks, such as keeping two traditional genes the right distance apart for other regulatory machinery to operate on them.

What counts as a gene?

Surprising new ways in which DNA can perform both its traditional and its new functions are being discovered. Genes can overlap one another or occur wholly inside other genes. Separate genes can be encoded in opposite directions in the same DNA molecule, one strand of the double helix coding for one gene product and the 'mirror image' on the other strand coding for another, quite different, gene product. A single gene can be read twice and the results pasted together so that some parts of the gene occur more than once in the gene product. Two traditional genes can be read together to produce a single, large RNA from which a third gene product is assembled. The products of DNA from widely separated parts of the genome are pasted together ('*trans*-spliced') to

produce a single 'gene product'. The term 'gene' can today refer to any sequence or sets of sequences that underlies one of the many tricks an organism can pull off using its genome and these various 'bags of tricks' can borrow parts from one another.

The reality of genome structure today challenges the conventional picture of the gene in the same way that the reality of particle physics challenges the traditional picture of matter. The 'particles' of the quantum world can lack such apparently essential features as having mass or being in some particular place. In the same way, just about any of the normal expectations we have when we hear the word 'gene' is violated by some important class of DNA sequences. Physicists changed their concept of a particle in response to the strange world that quantum physics revealed. Just so, in the 'post-genomic' world scientists continue to talk about 'genes' but often mean something quite at odds with the picture of the gene found in school textbooks.

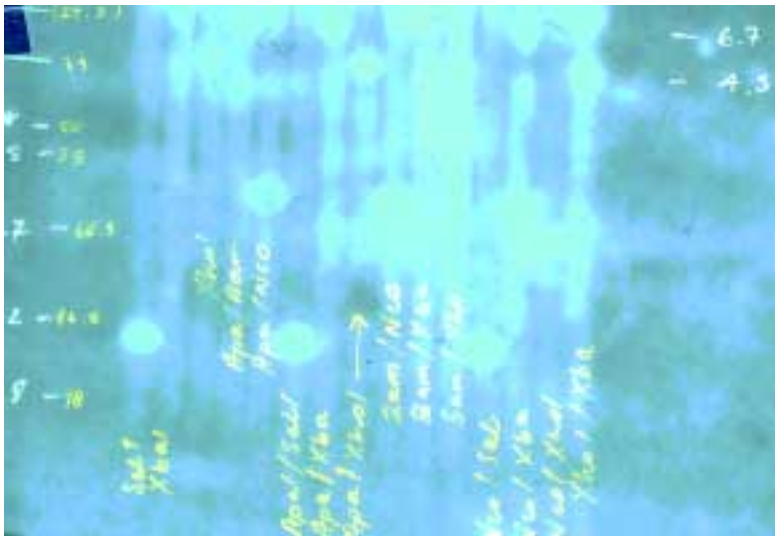
How many genes make a human?

These conceptual issues become highly practical ones when scientists 'annotate' a sequence of DNA. It is not at all obvious which sequences to count as genes. The public and the private Human Genome Project groups counted a similar number of genes, but significantly often these were not the same genes. The simplest product of annotation is an overall figure for the number of genes in a genome, such as the strikingly low figures produced in the immediate aftermath of the sequencing of the human genome, ranging from a conservative 26 000 to a generous 40 000. But simple gene counts disguise the highly problematic nature of most initial annotations. In our current state of knowledge many different approaches to identifying genes are defensible, and different research groups often produce only partially overlapping lists.

Representing genes

In order to get a better idea of how these complexities might impact on the real world of day-to-day biological or medical research, we are investigating the different usages of the term 'gene' in different fields of contemporary biology. The research employs a questionnaire in which biologists and practitioners from a range of research fields are asked to consider real cases in which some set of DNA sequences take part in the production of some gene product (a 'transcription event'). They are asked if the case in question involves one gene or more than one gene and to indicate the position(s) of each gene on the DNA. Finally, they are asked whether some other descriptions than 'gene' are needed to adequately label the DNA and whether critical information is missing from the description of the transcription event.

Our results should reveal whether and how the concept of a gene is changing as a result of developments in molecular biology. Is there a correlation between the kind of research a molecular biologist is doing and the gene concept that informs this work?



Are biologists moving from a gene-centered view to broader, whole-genome oriented approaches, by loosening the current conception of a 'gene' or by replacing it with several, more differentiated terms?

One important aim of our work is to put to the test the widespread view amongst commentators on contemporary biology that the existing concept of the gene is simply not up to the job. Did 'the century of the gene' end with the celebration of the millennium, as the leading MIT scholar Prof. Evelyn Fox-Keller has argued? Some commentators have suggested that the diversity in understanding of 'gene' in contemporary biology is so great that it leads to frequent miscommunication between researchers. If borne out, these suggestions might be of direct value to biologists. They would also have implications for the public understanding of genetics and for bioethics, fields which involve taking claims made in a specific research context and applying those claims to derive broader lessons for ethics and public policy.

The representing genes project is being carried out by the University of Pittsburgh in association with Egenis.

Karola Stotz works on the 'Representing Genes' project based at the University of Pittsburgh, and is a visiting fellow at Egenis.

attitudes to genomics

Public attitudes towards genomics

Richard Shepherd: principal investigator for the Attitudes to Genomics Survey

Recent advances in genomics have had a mixed reaction from the public and the success of future applications may depend on the public's perception and receptiveness. The Attitudes to Genomics Survey aims to provide a basic understanding of current attitudes among the general public towards genomics and how these attitudes are formed and change over time.

Advances in genomics in recent years have led to substantial developments in pharmaceuticals and medicine. However, applications in food and agriculture have met with negative public reactions in Europe and future medical applications may also present major issues of public concern. There is a clear need to understand how people think about and make decisions in this area. Against this background, the ESRC commissioned a major project, Attitudes to Genomics.

Initially, a national baseline survey was conducted on approximately 3,500 people as part of the 2003 British Social Attitudes Survey. This included some 60 genomics-related questions as well as questions on demographics and attitudes in other domains. The genomics questions covered knowledge, awareness/engagement, core values and beliefs, trust, general attitudes to genomics, history of genetic illness, use of genetic data, gene therapy, antenatal testing, human cloning and GM crops and foods. The fieldwork was completed between June and September 2003 and the data are currently being analysed. Results will be published as part of the British Social Attitudes book.

The survey work was complemented by an intervention study examining the effects of information provision using a sub-sample from the original survey. Of those in the sub-sample, one third received a short film on the science behind genomics, another third received the same film on the science, plus a section on regulation, and the remaining third formed a no intervention control group.

While the survey results give an indication of how people respond to short questions on aspects of genomics, the project also includes a series of six vignette studies which allow a more detailed analysis of how people respond to systematically varied short scenarios. These studies each have the same format with baseline measures of attitudes, ambivalence, trust and uncertainty, followed by a scenario designed to test different hypotheses in the different studies and then a further set of questions to test the impact of the scenario. This work is ongoing.

The qualitative work on the project comprises focus groups, interviews and textual analyses. There will be twenty focus groups: four with people affected by genetic diseases, four with people concerned about the environment and a further twelve groups with members of the general public, four focusing on genetic diseases, four focusing on environment and four on wider concerns. The responses will be analysed using discourse analysis.

The focus groups will be complemented by 30 interviews with key individuals in medicine, science, industry and lay interest groups. There will also be 60 interviews with members of the general public who have watched the information intervention films in order to understand more fully the responses from the quantitative intervention study.

The final part of the qualitative research is a textual and visual analysis of representations of genetic technologies in UK newspapers and on UK terrestrial, cable and satellite TV. The leaflets and websites of 20 identified 'key interest groups' concerned with issues related to genomics are also being sampled, as well as UK governmental informational texts published over the last two years. The latter category includes such texts as green and white papers, select committee reports, reports from executive agencies and non-departmental advisory bodies such as the Advisory Committee on Releases to the Environment (ACRE) and the textual content of departmental web-pages. Finally, the films used in the information intervention will also be subjected to textual and visual analysis. This work is ongoing.

Developments in genomics will affect many areas of life over the coming years. These will have major social and economic implications for society and the individual. Public responses to these technological developments will be key to whether the technologies are adopted and therefore we need to develop an understanding of how different sections of the public respond to the applications and the issues raised. The situation is very dynamic with beliefs, attitudes and behaviours likely to change over time and to vary between different types of applications and their relevance to the individual. Taken together, the studies in this project represent one of the most substantial investigations of attitudes towards genetic technologies in the UK and will provide an indication of how developments in this field might progress in the future.

innogen

Genetics and life assurance should we legislate?

Scientific progress in the field of genomics has been accompanied by broader social concerns regarding the use of clinical information. The policy framework around genetics and life assurance has become a particularly contested arena for public discussion in the UK. The concern that insurers might use new kinds of genetic test information to deny people cover has enjoyed broad coverage by the media, and a number of clinical, patient and consumer groups have painted dystopian futures of a socially and economically excluded 'genetic underclass' emerging in society. In the mid 1990s, the UK began a lengthy advisory and regulatory process on the issue of genetics and life assurance, largely in response to perceived public fears of genetic discrimination.

The outcome of the exercise was a five-year moratorium (due to end in October 2006), which prohibited UK insurance companies from using certain types of genetic information.

NO

The Insurance Industry's Perspective

James Mittra

The moratorium restricting the right of insurance companies to use certain kinds of genetic information legitimised the concern of various advocacy groups that 'unfair' discrimination against the 'genetically disadvantaged' was likely if restrictions were not imposed on corporate practices. The moratorium was essentially a temporary compromise between those who wished to see a permanent legislative ban on insurers' use of genetic information, and the commercial sector who did not wish to have their 'right to underwrite' permanently lost.

Social and commercial interests have been artificially polarised. Social interests have come to represent concern for the vulnerable and genetically at-risk. Commercial interests have come to represent crude economic rationality over social need. A number of powerful 'myths' articulated by various advocacy groups has helped drive the decision-making process in this direction.

There needs to be a broader debate on the nature of life assurance and its social role. Commercial insurance is essentially in the 'business of discrimination'. If an applicant represents a 'statistically significant' risk, it is only fair that the insurance premium reflect this. Insurers claim that such discrimination is 'fair' if it is based on sound actuarial evidence. 'Unfair' discrimination occurs only if the insurer misinterprets the data or allows value judgements to pollute the underwriting process.

From the insurer's perspective, the cause of an individual's risk is less important than the statistical consequences of it. Insurers defend the right to access applicants' medical records on the grounds of 'equity' (applicants pay a premium commensurate with their risk) and 'adverse selection' (at-risk applicants may purchase excessive insurance at standard rates if they are not compelled to disclose the information).

Many people have begun to challenge the principles of commercial

insurance by arguing that to deny insurance to individuals based on their 'genetic risk' is fundamentally 'unfair'. However, absent from the debate has been any consideration of the type of 'social good' life assurance represents, which is crucial to our understandings of social exclusion and the ethical limits of commercial insurance. If a particular form of insurance is a 'primary social good' (something people ought to have access to because society deems it essential), there may be justification for restricting commercial freedom or changing the system of provision. However, if an insurance product is considered a 'non-primary social good' (something that has a social value but is not considered essential), society may have less justification for imposing restrictions on the commercial sector. The debate in the UK has focused on life assurance, and talk of social and economic exclusion appears less convincing when one looks at the reality of life assurance provision. It is not a product to enable the 'poor and downtrodden'. Instead, it exists primarily as a means for the wealthy to protect their financial interests. If it is not a 'primary social good', one could argue that providers of life assurance have a legitimate right to use existing underwriting methods to classify applicants.

Certain kinds of genetic information are claimed to be 'special' and in need of unique regulatory attention. In the context of insurance, new information derived from DNA and biochemical testing is described as qualitatively different from existing forms of medical data, and its use to load insurance premiums presented as 'unfair' if it leads to unacceptable social exclusion and affects peoples' willingness to undergo genetic screening. However, new kinds of genetic tests will likely just refine the existing underwriting process. They may prove more accurate but the information itself is little different from family history, which has always been used by insurance companies. Critics of the insurance industry also often fail to recognise that genetic tests can rule people into insurance as well as out of it. In the absence of a genetic test, applicants with a family history of Huntington's Disease are charged a 50% loading on their life assurance premium to reflect the fact they have a 50% chance of developing the condition. However, if such applicants took the Huntington's gene test, half would not have the gene and be able to acquire standard rates. Sceptics rarely appreciate that it may be unfair to provide special protection to the 'genetically disadvantaged'. Granting protection only to applicants whose risk is discovered through a specific diagnostic test is unfair to other individuals in the insurance pool whose diagnosis is derived from a non-genetic test.

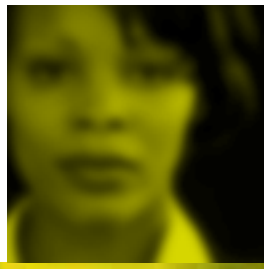
Critics have also ignored the fact that 95% of applicants for life assurance pay standard rates, 4% pay a loaded premium and only 1% are denied cover, suggesting that the life assurance industry is highly competitive and uses minimal underwriting. One should not assume that genetic testing will fundamentally affect the existing life assurance market.

Policymakers and the public have failed to recognise the 'creative capacity' of the insurance industry to respond flexibly to emerging technologies and markets. Special insurance products such as smokers annuities have enabled far more people to enjoy protection from risk, and new innovative insurance products will probably be developed in the unlikely event that the dystopian 'genetic underclass' vision becomes a reality.

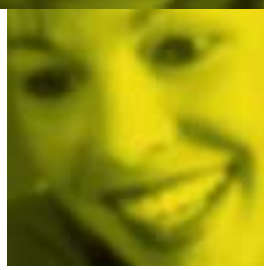
It is important in these controversial debates that we have a good understanding of industry perspectives, and the underlying reasons for them, so that when governments do regulate they have comprehensive and impartial academic research to support their judgements.

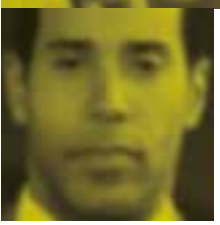
This article is based on evidence derived from a series of interviews, which included a number of representatives from the insurance industry, conducted as part of a three-year PhD thesis on the UK's policy and regulatory framework around the issue of genetic information and access to life assurance.

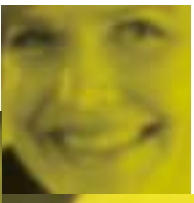
James Mittra is a research fellow at Innogen and can be contacted at ***james.mittra@ed.ac.uk***

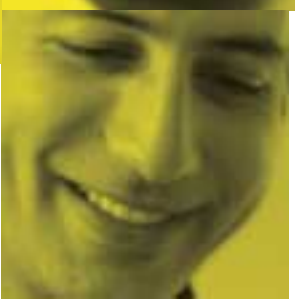










YES

John Sulston

James Mittra's article makes a number of important points, but the context in which he places them results in a flawed conclusion.

First, he is right to say that public discussion over genetic discrimination has been agitated. This has been true at every stage of the century-old struggle for genetic equity - the right of a woman to vote, the right of a black person to work, and the rights that are now being contested. The reason for the agitation is that only through free and open public debate, with all its flaws, do we attain social improvement.

He is also right to say that genetic information should not be regarded as exceptional. No aspect of a person should be taken as grounds for discrimination against them. The special point about genetic information is that the ability to collect personal genetic information has increased greatly of late, and will increase much more in the future, so it's something we have to think about now - but certainly not to the exclusion of other information.

With regard to life insurance, we are dealing with social change. The issue is whether it has now become the right of citizens to have levels of insurance sufficient to cover house purchase and protection of family income. Most of us agree with the proposition, since our means are modest enough that a mortgage is essential for the former, and our prudence is such that we hesitate to reproduce without the latter. If we democratically agree on these perceptions then we shall ensure through regulation that a modest level of insurance is available to all, as of right, in the same way as healthcare and unemployment benefit. In other words, life insurance has indeed become a 'primary social good' - with one proviso, that the sums assured reflect average needs. That is exactly what has been put in place by the existing voluntary (not statutory) moratorium on life insurance. Concerns about adverse selection - the possibility that people, knowing through personal genetic information that they have reduced life expectancy, benefit their estates by insuring for large sums - is taken care of by limiting the assured sum to £300K - 500K. Above that level, we can indeed regard insurance as a non-primary good, and so the moratorium does not apply.

The fact that life insurance cover is currently denied to only 1% of applicants, and the expectation that unregulated genetic testing would not greatly increase that number, is reassuring. But it is not a reason for inaction. On the contrary, it nullifies any concerns about cost. If, say, 2% of applicants are enabled to obtain cover when without regulation they would be denied it, they can all come on board without the rest of us noticing. It will make a huge difference to the 2%, though. A civilised society does not disregard the fate of its minorities.

So what is happening now? Exactly what Mittra calls for - a broad debate on the nature of life insurance and its social role. A section of the Human Genetics Commission is working on the topic, taking evidence from all interested parties. Their process includes regular meetings with the Genetics and Insurance Committee, whose remit is to evaluate the accuracy and relevance of genetic tests for insurance purposes.

However, the issue in discussing life insurance is not simply whether the views of all stakeholders have been balanced. If we do collectively sustain the view that modest insurance is a primary social good, this will not be an attack on the insurance industry for poor underwriting, as Mittra suggests, but merely a sensible adaptation to social progress. Finding a way to put that conclusion into practice will then be a matter for practical arrangements that use the 'creative capacity' of the industry.

Market forces alone cannot deliver social justice. That job is for democratic consensus, providing a framework in which market forces can operate benignly.

Sir John Sulston is the former director of the Sanger Centre, and won the Nobel Prize in Physiology of Medicine in 2002

Making the biosciences work for the poor

innogen

James Smith

New innovations in biotechnology could be used to alleviate a range of agricultural and health-related problems such as the HIV/AIDS epidemic and food security issues that are currently experienced in developing countries. So why is this not happening? What are the constraints on the utilization and dissemination of technologies that could transform lives? How can we address these problems and find practicable solutions?

It is commonly believed that new developments in the life sciences can be used to overcome major agriculture and health related problems, and that the biosciences can provide solutions to a range of problems in the developing world. Technologies such as genetically-engineered vaccines, for diseases such as HIV/AIDS and malaria, and the advent of genetically modified staple foods with enhanced nutritional value, disease and pest resistance such as rice, potatoes, corn and cassava could transform lives. However, this idealistic view does not take account of the pharmaceutical and agricultural companies' unwillingness to address the needs of the sickest and the poorest. Perhaps the most telling and stark statistic is what has become known as the '10/90 gap', where 90 per cent of all medical research is targeted at problems affecting only 10 per cent of the world's population.

At a recent conference in Belgium, Louise Fresco of the FAO stated that 'the gap between rich and poor farmers, between research priorities and needs, and between technology development and actual technology transfer, is widening', and endorsed the idea that biotechnology must be redirected to address the pressing needs of the poor. Poor countries and farmers should have access to genetic resources and to the technologies and means to use them. Public-private partnerships could play an important role in overcoming this 'molecular divide'. Innovative public-private partnerships in biotech research need to be encouraged in order to promote the transfer of technology to the public sphere while maintaining incentives for private-sector R&D.

Partnerships between research institutions are attempting to draw together innovative science with local experience of applying the science. Funding bodies and donors, too, are playing a role, placing increasing emphasis on poverty alleviation and development objectives through the development of new bioscience technologies.

Partnerships, however, tend to be served up as a solution to a range of disconnects that prevent the biosciences fully engaging with the developing world. Partnerships are seen as inherently good in both innovation systems and development; partnerships have been the driving force behind a shift from exogenous to endogenous technological development. Indeed, in many respects, partnerships literally are the innovation system. In reality, such partnerships are often nothing more than politically, economically and culturally constructed transfers of resources between institutions. The implied undercurrents of equality, strategy and common purpose are often singularly lacking, and in reality, partnerships provide superficial representations of inclusiveness, efficiency and egalitarianism.

Partnerships can also be seen as a way of linking innovation and development, and can thus become a battle ground, caught between, and bridging, two separate bodies of knowledge and the systems they represent.

Our recent research on tissue culture banana projects in Kenya indicates that projects can be judged successful, in terms of innovation theory, but a failure from the perspective of development. This project created and managed a complex network of partners, with a range of

institutional structures, from the north, the south, the public and the private sectors. However, the project has so far failed to achieve its own development outcomes because it generated policy prescriptions based on a series of false premises and poor analyses. It is unfortunately not simple to unstitch one domain from the other. The development process is inherently outward looking with little space for self-reflection or self-learning. The banana innovation system, because it had partnered with and within the development domain, was obliged to adapt itself to operate in that environment. The partnerships within the system end up articulating two movements: the tensions between the development and innovation domains, and the tangible flows of resource transactions that the partnerships focus and direct.

Health-focused partnerships appear to be different, however. Our work on the DfID-sponsored East Coast Fever project and on the International Aids Vaccine Initiative (IAVI) indicates that these more tightly focused, results-oriented partnerships are more successful in finding efficiencies in the research and dissemination of technologies precisely because they seek to avoid issues of 'development'.

To transform bioscience advances into benefits for the poorest and most vulnerable, we must understand the internal dynamics within partnerships. A culturally embedded critical reading of partnerships can map these dynamics and foster more focused, coherent and relevant partnerships for innovation and development. The culture of IAVI shaped by a very corporatist, private sector vision of how to drive research is markedly different from the panoply of small NGOs who are increasingly driving agricultural biotechnology dissemination in Africa. Our research is attempting to understand the complex, iterative dialectic inherent in such partnerships and to pick out which policies, philosophies and organisational strategies are producing tangible results, and which are not.

James Smith is a research fellow at Innogen and can be contacted at ***james.smith@ed.ac.uk***

Too much genetic information?

arguing the case for and against non-therapeutic genetic testing in childhood

Susan Hogben and Paula Boddington

Genetic information has come to be a prestigious, desirable and difficult-to-resist 21st century artefact. Yet debate still abounds regarding the production and distribution of genetic information; what sort of genetic information is appropriate; when ought it to be generated and to whom should it be made available?

These concerns are further complicated when we consider childhood genetic testing (CGT) and especially the judiciousness of testing which will not provide any immediate medical benefits to the child. Such situations include genetic testing which may reveal that a child is a carrier of a condition which will not affect them but may have consequences for any future generations (so-called carrier testing) and genetic testing for adult-onset conditions which reveals information about the future disease status of a child (so-called predictive testing).

The benefits of non-therapeutic genetic testing are something many medical professionals, ethicists and parents can often only speculate about. Something which is complicated by the many ethical issues involved in childhood genetic testing; What are the rights and responsibilities of parents? When does a child cease to be a child? Does respect for autonomy mean that the child must make decisions for herself, or that in order to be autonomous, a child should have access to as much information as possible including information about her genetic status? What should be the roles of professionals as gatekeepers to genetic services? Whilst each of these issues and others demonstrate the complexity of reaching conclusions about the appropriateness of genetic testing in childhood, conclusions and recommendations are indeed drawn.

Our preliminary research examining policy documents produced by geneticists, paediatricians and patient advocate groups from the UK, USA and Canada reveals interesting differences in attitudes to non-therapeutic genetic testing. Whilst all the position papers strongly endorse delaying predictive testing until the child is competent enough to decide for herself a number of the papers fall short of ruling out testing for carrier status to a similar degree. So how does this difference come about? There seem to be a number of complementary factors that influence why the recommendations differ.

Carrier and predictive testing: a distinction that makes an ethical difference

The position statements consistently maintain a strong distinction between carrier and predictive testing by assigning the information derived from genetic testing differing degrees of severity. Information arising from predictive testing is presented as being more serious than information about one's carrier status even though predictive testing is also at the same time carrier testing, and despite the fact that carriers of some conditions may also manifest some symptoms. The seriousness or burdensome nature of genetic information arising from predictive testing results from it being about an individual's own disease status. Interestingly, the onerous nature of this information is frequently dependent on the use of Huntington's disease (a neurologically debilitating condition that is untreatable, fatal and generally regarded as devastating in its impact) as the exemplar for adult-onset disorders and also for predictive testing. By consistently presenting the consequences of predictive testing as highly undesirable carrier testing comes to be viewed as 'less serious' 'of minimal risk', important merely in reproductive decision making, of purely reproductive significance to the child in the future. So it is assumed, in a kind of genetic egoism, that an individual will be more concerned about her own disease status than that of any possible children or grandchildren.

It seems that the ethical arguments which protect children from predictive testing rely strongly on respect for autonomy, whereas the arguments which render carrier testing less serious tend to be influenced by debates around potential harms and benefits. Presenting carrier testing as less serious opens the doors to parental influence in decision-making and might enable practitioners to permit childhood genetic testing for carrier status in situations where predictive testing would be frowned on. Of course, this too will be influenced by the particular character of the disease such as the age of onset, potential interventions, progression etc.

Whilst the recommendations reveal much about the ethical complexity of non-therapeutic testing the practical consequences of childhood genetic testing are still unclear.

Our research examines these issues from two perspectives. By interviewing families where genetic testing is a possibility we are investigating why parents consider it useful to test a child, if and how children are involved in the decision making process and what children themselves say about making a decision to be tested. We are also exploring the issue retrospectively by asking people who were tested in childhood or were, say, identified as carriers of a genetic condition pre-natally, how they feel about having this information and what effect, if any, it has had on their life choices, their relationship with siblings, other family members, friends and so on.

Susan Hogben and Paula Boddington are research associates at Cesagen. If you'd like more information on any aspects of the project mentioned above please contact Dr. Susan Hogben Cesagen, Cardiff 02920 870026 or hogbens@cardiff.ac.uk or visit the Cesagen website at www.cesagen.lancs.ac.uk.

The internet as a patient information tool

egenis

Paula Saukko

Resorting to the internet to obtain information on health is becoming increasingly popular. People with genetic conditions are particularly likely to surf for knowledge and contact others with similar issues through the net, as these conditions are often rare, new and poorly understood. Based on our research on an on-line discussion group for people with genetic thrombophilia it seems the conversational nature of the net may provide a unique tool to help people make sense of a complex genomic condition. However, the net may also feed people's interest in problematic treatments.

What's been said before?

Research on internet and health has often focused on the accuracy of information on the net. Some have noted that on-line patient discussion groups provide emotional support and validate concerns, such as dissatisfaction with treatment, that do not often get a hearing. In our research we looked at a net-forum for people with thrombophilia, or a susceptibility to deep vein thrombosis. We analysed the content of the conversation (what is being discussed) as well as the form of discussion (how is it being discussed). Our aim was to explore what is unique about the internet as a patient information tool.

Thrombophilia: our case study

Thrombophilia is a common, complex genetic condition. This means that about one in twenty individuals in Caucasian populations have one of the genetic alterations that increase, even if only slightly, an individual's chance to develop clots in their deep veins. There are several genetic markers that have been shown to increase an individual's susceptibility to deep vein thrombosis. A genetic test, that scans for these markers, is now in use in the UK. Furthermore, many environmental factors, such as oral contraceptives, hormone replacement therapy, surgery, pregnancy, immobility and smoking, increase the risk of developing clots.

The genetic test for thrombophilia is useful for people who are diagnosed as being at risk. The test allows them to take measures to prevent clots. Such measures might include not taking oestrogen, changing lifestyle, and taking prophylactic medications during high-risk situations. However, medications may have serious side-effects. This multi-dimensionality of the condition creates a challenge for health communication.

The internet discussion group www.fvleiden.org

The discussion group website that we studied has about 1000 subscribers, many of them women from North America. The list is associated with informational web-pages and the moderator of the site and some members are active in a newly formed patient advocacy-group. The list is rather lively, and the traffic in 2003 was around 25 posts a day. We analysed 3600 posts exchanged during a six-month period in 2003 and

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What was interesting about the discussion on anti-coagulants was the way in which it could flesh out many perspectives on the topic. Traditionally health communication has been viewed in terms of a doctor or experts relaying information to patients or the general public. Internet discussion-groups, however, simultaneously bring together different contrasting and complementing perspectives. Thus, the exchanges on anti-coagulants could, for example, argue that taking Aspirin was fine, as even if it was not an effective prophylactic it was not harmful like Warfarin, whereas others would point out that Aspirin could be harmful, and still others would note that they felt betrayed when they clotted on Aspirin and were not put on Warfarin. All these different views were feasible, and the internet as a medium allowed the participants to explore the different possible actions and outcomes associated with a complex condition, such as thrombophilia where there are no hard and fast answers to most questions.

However, the fact that anti-coagulants were the most common topic also raises the question of whether the net feeds people's interest in treatments that may be problematic. An individual who has inherited one of the alleles associated with thrombophilia and has not had an episode of deep vein thrombosis, has no reason to use anti-coagulants, as the risk of potentially fatal internal bleeding associated with the drugs is more dangerous than clotting. Research on public and

EGENIS

Seminar Series

Egenis runs weekly seminars at the University of Exeter. Details of forthcoming seminars are available on the website. **www.exeter.ac.uk/egenis**

British Association Festival of Science, 4-12 Sept 2004

This roving festival attracts over 350 scientists annually and boasts over 100 open access talks, workshops and events. Egenis staff have been awarded a consultancy to co-ordinate the *Festival in the City*. Over 40 events for local people include the RAF falcons parachute teams leaping from the skies, night time star gazing at the observatory, a public meeting on GM and all night viewings of sci-fi films. In addition the directors of all three Genomics Centres together with Dr Phyllis Starkey MP will present a session on objectivity and expertise in science hosted by Egenis director, Professor John Dupré, who is president of the BA's history of science section.

Café Scientifique

Egenis organises a series of free lectures and discussions at the Phoenix Arts Centre as part of the Café Scientifique network. This venture is independently funded by the University of Exeter's community fund and sponsored by the Met Office. The café is a place where a lay audience can meet with professionals and scientists from various areas of expertise. Professor Robert Winston will give a talk in September on assisted reproduction. **www.ex.ac.uk/cafescientifique**

Representing Genes

Results of the Representing Genes project will be announced at a media friendly seminar on 13th-14th May 2005. Attendance is by invitation only.

2005 Conference

The Egenis conference 'Genomics in Context' will take place on 29th and 30th September 2005. Genes are now seen as a part of an interactive network that encompasses the genome, the cell, the organism and the natural and social environment. The conference will examine the implications of this in health, agriculture and more generally.

CESAGEN

Seminar series

Cesagen runs regular seminars at Lancaster University which are open to all. Details can be found on the Cesagen website. **www.cesagen.lancs.ac.uk**

Genetics and the Natural workshops

Cesagen will be running two workshops on the theme of genetics and the natural. Speakers at the first workshop on September 17th will include Kate Soper, Finn Bowring, Bron Szerszynski and Alan Holland. The second workshop on April 21st 2005 will include contributions from Tim Hayward, David Cooper and Tom Baldwin. Both workshops will look at the distinction between the natural and the unnatural and consider its implications for genetic modification and other technologies.

2005 2nd International Conference

Cesagen will be holding its 2nd International Conference on the theme of Genomics and Society at The Royal Society in London on April 12th-14th 2005. Topics covered will include genomics and it's implications in: the transformation of knowledge production; economics and innovation; ethics and regulation; identity and social organisation; risk and responsibility; global discourse and cultural capital. The conference will also address Cesagen's cross-cutting themes of globalisation, health, environment and public engagement. A call for papers is now available on the website. For further details visit the conference website at **www.cesagen.lancs.ac.uk**

INNOGEN

Innogen Annual Conference

Innogen would like to announce the Evolution of the Life Science Industries conference to be held at the Edinburgh International Conference Centre on 23rd-25th February 2005.

The ELSIS Conference will be unique in bringing together senior managers from industry, scientists, policy makers at national and international levels, and academics contributing in a practical way to improved policy making and improved strategic decision making in companies, regulatory agencies and government departments.
An important aim of the conference will be to promote constructive dialogue among these diverse participants and to contribute constructively to the shaping of future pathways of innovation in the life sciences.

This major international event will cater for up to 500 delegates. Topics to be covered in these sessions will include:

- the future of 'big pharma'
- pharmaco-genomics and industry strategies
- stem cells and cloning – their potential impacts on therapies
- genetic databases – impacts on public health
- evolution of life science industries in developing countries
- policies for promotion of the life sciences
- policies for guidance of the direction of innovation and for its regulation
- the impact of ethical and stakeholder pressure on innovation
- communicating about life science-related innovations

Seminar Series

Innogen runs monthly open seminars at the University of Edinburgh or The Open University. Please see **www.innogen.ac.uk** for details of subsequent seminars.


ESRC
genomics
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E·S·R·C
ECONOMIC
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RESEARCH
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