



# **Biobanking in Aotearoa New Zealand:**

A case study of  
the New Zealand Rare Disease Biobank  
operated by the NZ Institute for Rare Disease  
Research Ltd.

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## **Abstract**

In the 21<sup>st</sup> century there has been a significant expansion in the knowledge available about human genes and their relationship to human health and illness. The completion of the Human Genome Project, developments in the digital processing of large bodies of genetic information and the availability of equipment that enables the visualization of cells at the molecular and sub-molecular level have generated escalating interest in the collection and analysis of samples of human and animal tissue. While systematic repositories of biological tissue have been a core resource for scientific work for many years, there is now increased interest in the establishment of tissue collections from which DNA can be derived and the possible genetic analysis of samples collected for other purposes.

These repositories of tissue are a vital resource for public good scientific researchers and commercial biotech companies. Increasingly they are referred to as 'biobanks' – resources in which communities and nation states and sets of nation states 'invest' as a component of 'the knowledge economy'. Some of these biobanks are population and public health focused – directed at identifying the interactions among genes, lifestyle factors (such as smoking and diet) and the impact of different social and physical environments. Other biobanks are more specialized and directed at creating tissue repositories associated with particular disorders. While DNA analysis may be component of the use of these repositories, they are also used to derive cell lines, explore cell responses to particular agents, develop diagnostic tests and inform innovation in treatment.

This report provides an overview of an emerging biobanking initiative – the NZ Rare Disease Biobank. This biobank is owned and operated by the NZ Institute for Rare Disease Research Ltd, a charitable company owned by the New Zealand Organisation for Rare Disorders (NZORD). The goal of the biobank is to encourage research relating to rare diseases in Aotearoa New Zealand through facilitating the collection of new tissue samples and the systematic documentation of collections of animal tissue relevant for the study of rare human genetic disorders. This initiative involves collating information about the available animal models relating to rare diseases generated by researchers in a range of different research centres and will eventually involve the collection and systematic storage of human tissue from people with rare disorders and their families. This New Zealand biobanking experiment is set in the context of international trends in the development of tissue banks, particularly patient-advocacy group involvement with the establishment of biobanks.

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## Setting the context – international biobanking initiatives

Over the last 30 years there has been an expansion internationally in biobanks – repositories of tissue that link genetic information and details about individuals' personal medical histories (Rose, 2001; World Health Organisation 2002; Einsiedel 2003; Tutton 2004; Williams and Schroeder 2004; Fletcher 2004; Busby 2006a, 2006b). However, as Petersen (2005: 275) indicates, 'biobanking' in this sense is not particularly novel. Clinicians and researchers have been assembling databases that include tissues and medical records relating to particular conditions for many years. For example, the Centers for Disease Control in the USA have been doing national surveys on health and nutrition for the last 30 years. These surveys include obtaining samples of blood, serum and urine that have been used for DNA analysis.<sup>1</sup>

In the last ten years, however, there has been increased interest in the establishment of population-based genetic databases which combine information about DNA and medical records. These databases are increasingly referred to using the metaphor of the 'bank' and a discourse of investment. A range of discursive repertoires<sup>2</sup> are exhibited by those seeking to recruit donors for such tissue repositories. These repertoires variously define participation in terms of 'altruism', 'gift-giving', 'genetic solidarity' and, in the case of patient-led biobanking, 'control' and 'management' of their own DNA.<sup>3</sup>

Biobanks,<sup>4</sup> or biomedical collections consisting of genetic data, other cellular material and medical records are increasingly framed by their advocates (scientists, patients advocacy groups, state actors, research institutes and commercial biotech companies) as 'investments' in the generation of new knowledge about the interactions between genes, social and physical environments, life style and health outcomes. They are a key component of 'the knowledge economy' - 'banks' of knowledge from which information can be generated for the better treatment of those with common illnesses like cancer,

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<sup>1</sup> See [http://www.cdc.gov/nceh/dls/dna\\_bank.htm](http://www.cdc.gov/nceh/dls/dna_bank.htm)

<sup>2</sup> Discursive repertoires are linguistic tools that are available to people as they construct talk about a particular topic. These repertoires are deployed by individuals, but are socially constructed – shared sets of understandings which are utilised in particular contexts. Discourse analysis involves attention to the way in which individuals and sets of people strategically use a range of shared understandings to interpret their environments. A key idea associated with attention to discursive repertoires is the idea that, while the understandings used are not unique to individuals, there is flexibility in their use and people may combine different repertoires as they encounter new situations that require response and analysis (Wetherall and Potter, 1992).

<sup>3</sup> See Tutton (2007); Tutton (2004); Tutton (2002); Petersen (2005); Human Genetic Commission (2002) and the Genetic Alliances BioBank website <http://www.biobank.org/> for discussion of these repertoires and examples of them in use.

<sup>4</sup> A biobank has been defined as: "any non-profit service unit that is aimed at the collection and storage of human genetic materials and the respective clinical data for a given period, or else for an indefinite period in the case of anonymous data, and is used for the purposes of diagnosis, study and research." (Pizzetti 2005: 1)

diabetes and heart disease, as well as genetic disorders. The most ambitious of these biobanking initiatives is UK Biobank which was established in 2002 and began recruiting half a million volunteer donors in 2006. The website for UK Biobank refers to the tissue and information database that is being established as a 'national treasure trove'.<sup>5</sup>

Against the background of the UK Biobank initiative and discussion about this collaboration between the UK Department of Health, the Medical Council and the Wellcome Trust, talk about biobanking was included in an experiment in public engagement with issues relating to genetic testing and biobanking during the first phase of the Constructive Conversations/ Kōrero Whakaaetanga research programme (2003-5). In the first phase of this programme 25 focus groups were conducted in different parts of Aotearoa New Zealand with members of community groups, whanau/extended family groups and other informal social networks which explored their responses to issues relating to newborn genetic profiling, direct to consumer genetic testing and the possibility of a hypothetical 'NZ Biobank' which relied on voluntary participation by people over 45.<sup>6</sup> The hypothetical NZ Biobank was based on planning for the UK Biobank. The fictive biobank used to facilitate discussion in focus groups was to be run by an organisation resembling a Crown Research Institute.<sup>7</sup> A fictional pamphlet aimed at prospective donors to this biobank was the catalyst for discussion.<sup>8</sup>

In the course of doing this research, information about an initiative to establish what was referred to as "New Zealand Rare Disease Biobank" became available. This biobank was very different from the UK Biobank initiative that had provided the model for the public engagement initiative associated with Phase One of the Constructive Conversations Research Programme. This research report provides an overview of this biobank initiative.

New Zealand Rare Disease Biobank is innovative in its intention to construct a data bank that includes both animal and human tissue. By definition, the diseases that are the focus of attention in this case are rare in humans;<sup>9</sup> consequently there are significant limitations on the number of human samples available for analysis, particularly in Aotearoa New Zealand with a population of 4 million people. However, a number of human genetic diseases are manifest in other animals (e.g. cows, sheep, cats and dogs). For this reason, animal samples and animals models relating to rare human genetic diseases are significant for the identification and treatment of human rare diseases. Aotearoa New Zealand

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<sup>5</sup> See <http://www.ukbiobank.ac.uk> Why we need your support

<sup>6</sup> See Hipkins (2004) for a discussion of the research methodology used in this study.

<sup>7</sup> See research materials used in first round of contact groups in the Constructive Conversations/Korero Whakaaetanga research programme

<http://www.conversations.canterbury.ac.nz/PHASEONE/overview.meetings.htm> and (Hipkins, 2004) for discussion of the methodology used in this research

<sup>8</sup> See Scott (2005) et al for a discussion of some of the ethical issues relating to biobanking that were articulated by the focus groups that participated in this study.

<sup>9</sup> Rare diseases are diseases identified as affecting less than one person in 2,000.

has a history of high quality veterinary research as an outcome of the significance of primary industry in the New Zealand economy. Knowledge about the importance of animal models of rare diseases produced by veterinary researchers in this country contributed to the idea of a virtual network of animal and human biological samples and associated data.<sup>10</sup>

The NZ Rare Disease Biobank initiative is still in a formative stage. Animal models relating to rare diseases have been identified and the features of these data resources and their location have been recorded and made available on a website associated with this biobank.<sup>11</sup> The plan is to develop the systematic storage of human tissue from those with rare genetic disorders in Aotearoa/ New Zealand and their family members. However, while some samples have been voluntarily donated and stored, the protocols for setting up processes of consent for the donation and use of human samples are still being developed.<sup>12</sup>

This rare diseases biobanking initiative complements other biobanks set up in New Zealand in the last twenty years, for example, the Cancer Society Tissue Bank (based in Christchurch), the Eye Bank (based in the Department of Ophthalmology, University of Auckland) and the Neurological Foundation's Human Brain Bank (based in the Department of Anatomy and Radiology, University of Auckland).<sup>13</sup> The Christchurch Tissue Bank is a central repository for donated cancer tissues used in genomic and proteomic studies which includes samples from over 2,000 donors. Most donors have given consent for researchers to access their medical records (99.6%), to send tissue out of the country (98.3%) and to the use of tissue by commercial collaborators (97.4%).<sup>14</sup> The NZ Rare Disease Biobank differs from these other tissue repositories in the extent to which it is driven by patient advocacy groups and citizen involvement rather than doctor and research scientist interest in storing human tissue for research purposes. All these initiatives, however, are underpinned by significant community-based fundraising.

Nikolas Rose (2007: 131) has argued that a new kind of citizenship is emerging against the background of development in biomedicine, biotechnology and genetic science. He refers to this as 'biological citizenship'. Enterprising citizens in the contemporary context must exercise not only community responsibility, but

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<sup>10</sup> The conception of this biobank and the factors that contributed to its development are discussed later in this report.

<sup>11</sup> See <http://www.nzordbiobank.co.nz>

<sup>12</sup> This report draws on a number of different sources of information, but particularly interviews with John Forman, the Executive Director of NZORD and the founder of NZ Rare Disease Biobank, whose availability for these interviews and detailed feedback on this report is gratefully acknowledged.

<sup>13</sup> See the following websites for information about these other biobanking initiatives:

<http://www.cancersouth.org.nz/tissuebank.shtml>

<http://ophthalmology.auckland.ac.nz/eyebank/eyebank1.html>

<http://www.neurological.org.nz/html/brainbank.php>

<sup>14</sup> See *New Zealand Medical Journal* 31 March 2006, 119 (1231): 12

<http://www.nzma.org.nz/journal/119-1231/1922/>

also “corporeal” and “genetic” responsibility at both an individual and collective level (Rose, 2007: 134). The exercise of what Paul Rabinow (1996) has referred to as ‘biosociality’ - forms of collectivity established on the basis of some shared biological status - precedes current research on rare genetic disorders, but takes particular forms as this research develops. Plans to develop a rare diseases biobank constitute an example of a form of biosociality and a “from below” form of biological citizenship in the New Zealand context (Rose, 2007: 142). In setting the context for discussion of the NZ Rare Disease Biobanking initiative, this section of the report explores the differences between population-based biobanking initiatives and disease specific repositories those that involve patient support/advocacy groups.

### **Population-based biobanking – the case of UK Biobank**

The population and public health focused UK Biobank was initiated in June 1999 as a result of interactions between the Wellcome Trust and the UK Medical Council. By 2002 it had become UK Biobank Ltd and funding had been organized from the WellcomeTrust, the Medical Council and the Department of Health. Currently this funding stands at £61 million. The Scottish Executive has become the fourth partner in this initiative and people in Scotland will also be selected to participate. The goal is to study the ways in which genes, lifestyle and environment affect people’s health. Advocates of UK Biobank state that this information will improve the prevention, diagnosis and treatment of a range of illnesses that particularly affect older people such as cancer, heart disease, diabetes, dementia and joint problems.

Recruitment of half a million people aged 40-69 began in March 2006 (initially in the Manchester region) and will continue for four years. Participants will be included in the study for up to 30 years. As they enter the study, donors provide blood and urine samples and they will have a health check, fill out a questionnaire and consent to their medical records being accessed as part of the data set. All tissues and medical records will be linked, but anonymous. With the exception of information about the outcome of the health check, information about individuals acquired for the database will not be given back to them or used in their treatment. Broad consent is sought to the use of their blood and urine samples and associated information about lifestyle and health status for public good and commercial research. Some participants may be followed up at a later date and asked to provide more information for the biobank. There will be no obligation to participate in these follow-up activities.

Analysis by social scientists of biobanking has tended to focus on issues relating to constructing trust on the part of ‘the public’ and the development of protocols with respect to privacy and informed consent (Wolpe 1998; Anderlik and Rothstein 2001; Annas 2001; Gottweis 2002; Tutton et al 2004; Kaye 2004; Petersen 2005; Busby 2006a). As Petersen (2005: 272) indicates, research on

public attitudes towards science has indicated positive responses to the potential benefits of science and technology, but low levels of confidence in their regulation (Levitt and Weldon 2005; Constructive Conversations 2005:26). There have been particularly negative responses from different publics to the commercialization of genetic information (Rose, 2001; Einsiedel 2003: 15-17; Du Plessis et al 2004: 24-5). In the context of this skepticism, those seeking to promote public involvement in initiatives like UK Biobank have worked hard to construct the project as an opportunity to exercise altruistic 'genetic' citizenship (Human Genetics Commission 2002: 7; Heath et al 2004) by donating tissue, completing questionnaires and allowing access to medical records. The UK Biobank website, now devoted to recruitment of participants in this database, presents individuals who provide their personal testimonies as to why they support UK Biobank.<sup>15</sup> It also invites participants in the study to help scientists "unlock the secrets of disease that will bring a better life for all". It focuses on how those participating will not be the immediate beneficiaries. Those who will benefit will be "our children and their children".<sup>16</sup>

The acquisition and storage of genetic information needs to be conducted with extreme care and must be governed by careful regulation (Human Genetic Commission 2002; UNESCO 2004). Issues relating to informed consent are articulated whenever there is public deliberation on the storage of genetic data. People feel strongly that those who have donated tissue should be consulted if it is to be used more than once for purposes not indicated at the time of donation. Participants in public discussions about biobanks generally consider that stricter rules should apply with respect to accessing genetic information than other sorts of medical information (Einsiedel 2003: 10-11).

Alan Petersen (2005) has analysed a series of documents produced between 1999 and 2004 to identify the discursive repertoires used by those communicating with citizens and potential participants about UK Biobank. He suggests that over that period of time 'consultation' and 'public engagement' tended to be used as strategies to construct this initiative as one that was democratic and attentive to the concerns of 'the public'. Mairi Levitt (2005) has looked critically at these 'democratic' aspects of public consultation on UK Biobank. She argues that, while from the start there were attempts to solicit opinion from different publics using a variety of different strategies, citizens were not given an opportunity to have an impact on the priority that might be given to commercial uses of the database versus public good uses. She is critical of the extent to which members of the public will be able to control the extent to which the biobank is used 'in the public interest'.

There was little response to criticism that UK Biobank would emphasize the genetic determinants of health at the expense of social factors because, while life

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<sup>15</sup> See UK Biobank website <http://www.ukbiobank.ac.uk> Why is it important that I take part? Participants' stories

<sup>16</sup> Ibid



style and environmental information was obtained, the genetic information was the only 'hard' data that would be available to analysts (Petersen, 2005: 281). Tutton (2007) has also reported on the range of discursive repertoires used by focus group participants in discussions about UK Biobank in which tropes such as 'altruism', 'expertise' and 'empowerment' feature strongly. He draws attention to the different forms of 'participation' offered with respect to biobanking – participation through providing research materials for scientists and participation in the governance of these data resources.

GeneWatch UK, an independent policy research group, has consistently adopted a critical position on the establishment of UK Biobank. Helen Wallace, Deputy Director of GeneWatch, has argued that UK Biobank is based on a fundamentally erroneous understanding of the place of genes in determining illness relative to environmental factors.<sup>17</sup> She asserts that rising levels of obesity and diabetes are not the outcome of an outbreak of 'bad genes', but by social environmental and lifestyle factors. Non-genetic factors have much more impact on the distribution of health and illness than genes. She indicates that the processes of environmental and genetic determination are infinitely more complex than the relationships presented by UK Biobank advocates (Watts, 2006). GeneWatch UK are concerned about the continued lack of clarity about the relationship commercial biotech companies will have to this biomedical resource. It has cautioned donors to be aware that pharmaceutical companies are interested in using this database to develop new drugs and will patent the knowledge they produce using this resource.

Scientists have not always agreed about the scientific merit of UK Biobank (Petersen, 2005: 278). While many scientists are enthusiastic about the biobank as a resource for research, some scientists have argued that it is a costly endeavour pursued at the expense of other forms of medical research. In the face of these criticisms, advocates of UK Biobank have consistently argued that the UK needs to embark on this world-leading initiative. Population-based research data bases depend significantly on a well-organized, centralized public health service – an advantage researchers in the UK have over those in the USA – their key competitors in biotech innovation in the health sciences (Petersen, 2005: 280).

The health gains for citizens have been a prominent component of publicity about UK Biobank. However, from the start it was also promoted as a resource for commercial companies and an income generating component of the knowledge economy. As Helen Busby (2006: 858) indicates: "discussions about genetic research in the UK have taken place against the backdrop of shifting relationships between commerce and the public sector". Asserting the need to protect human tissue from being defined as a commodity, while recognizing commercial interest in accessing this tissue and developing patentable products

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<sup>17</sup> See statement made by Helen Wallace in 2006 as UK Biobank began to recruit participants. <http://www.genewatch.org/>

from the knowledge generated through it, has been a key concern of various ethical committees and commissions over the last decade (Tutton, 2004; Busby, 2006: 859).

Research in the UK and elsewhere, including New Zealand, has indicated that citizens are very skeptical of the motives and actions of commercial biotech companies and especially the pharmaceutical industry (Einsiedel, 2003; Du Plessis et al 2004; Petersen 2005). However, state health actors are very conscious that the availability of drug treatment usually depends on development of products by pharmaceutical companies that depend on state investment in basic and applied scientific research. Pharmaceutical companies will be able to pay for access to UK Biobank data, subject to the research they want to pursue being passed by ethics committees and being consistent with the goals of UK Biobank. The payment by companies for access to this database will be an important stream of revenue for UK Biobank. At the same time, independent structures have been set up with respect to ethical practice which should militate against the economic power of the pharmaceutical industry with respect to the use of this tissue repository. An independent UK Biobank Ethics and Governance Council was established in 2004 that includes as its brief the consideration of issues relating to the commercial use of the biobank. It has responsibility to consider the rights of research participants and “the general public” and ensure that the Ethics and Governance Framework established in 2003 is observed.

### **Disease specific biobanking initiatives - The case of EuroBioBank**

UK Biobank was preceded by well established disease-specific repositories of tissue and genetic information. The donors to these data resources were patients presenting with particular disorders and sometimes members of their families. Participants were approached not through strategic sampling of GP patient lists (as is the case with UK Biobank), but through clinicians involved in treatment who invited patients and their families to allow blood and other tissue used for diagnostic purposes to be used for research (Busby, 2006a: 853). These databases – as much ‘biobanks’ as the UK Biobank since they combine tissue samples and medical records – have attracted less attention than the population-based UK Biobank. They are nevertheless significant sources of bioinformation. Research participants may have much clearer understandings about what sort of research is needed and why scientists are interested in their donated tissue and access to their medical records. On the other hand, requests for tissue samples usually occur in treatment situations in which it may be difficult to refuse such requests. Recruitment of people to participate in disease-specific biobanks is therefore both more straightforward and potentially more complicated than population-based biobanking.

In the last decade, attempts have been made to integrate these disease-specific databases and establish networks of connection among different tissue

repositories. A key example of this is EuroBioBank – the first network of biological databases to be set up in Europe. EuroBioBank was established in 2003, and articulates as its core goal the integration of information about “a critical mass of collections” and progressing research on rare diseases. In the context of an enlarged Europe, those affected by rare disorders are estimated as approximately 20,000 people.<sup>18</sup> An expanded Europe and the European Fifth Framework Programme “Quality of Life and Management of Living Resources” provide a context for claims for resources to set up a network of biobanks for rare diseases and the development of ethical protocols to be used in the collection of any new samples for databases to be included in this network. Significantly, EuroBioBank uses the same ‘banking’ metaphor adopted for UK Biobank. However, the focus is different, since attention has been directed primarily on the collation and integration of information about rare diseases databases.

EuroBioBank is directed at developing “high quality criteria for common banking practices” that are relevant to the different types of material collected (e.g. blood or other tissue) and the construction of a centralized database and website that will present the collections and facilitate researchers’ access to these collections. It is informed by the idea that a biobank does not have to be a single repository of tissue, but can be a mechanism for sharing data across sample collections in different locations. A key focus is the development of best-practice with respect to informed consent, storage, organisation and categorization as well as the development of cell culture models as tools for research. This network of databases is consistent with the NZ Rare Disease Biobank initiative which is also conceived of as a network of databases directed at facilitating research on rare diseases. The EuroBioBank initiative is, however, entirely a human tissue database. It does not involve the human tissue/animal tissue hybrid network which is the vision for the NZ Rare Disease Biobank.

Work is proceeding on arriving at consistent procedures with respect to DNA, tissue and cell culture preparation for EuroBioBank as well as ethical protocols consistent with European Union guidelines. By August 2005, all the partners’ collections were available on the website (including 140 cell collections and 543 DNA collections as well as some tissue collections from rare diseases patients). Researchers who currently want to use samples listed on the website can access a form requesting the use of particular research material. These samples are then accessed much more rapidly than would otherwise be the case. At this stage of its development, the biobank consists of 16 founding partners from 8 EU countries and includes 12 separate biobanks.

According to Majumder (2005) virtual databases are increasingly being developed which involve creating networks across institutions using the Internet for communication between these institutions and between them and others. Samples are digitalized and access to them maximized through the use of digital technologies. Majumder considers a number of different cases of virtual

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<sup>18</sup>See EuroBioBank website - [http://www.eurobiobank.org/en/information/info\\_institut.htm](http://www.eurobiobank.org/en/information/info_institut.htm)

databases and raises some issues about how consent is managed in the context of virtual databases. She argues that the best informed consent processes entail research specific informed consent, rather than one off consent for the use of samples for any research purpose. She suggests that digital technology can be used to enable donors to control the use of their tissue samples after donation.

What were the catalysts for the development of EuroBioBank? One factor cited as precipitating this initiative was a consistent flow of letters from patients and their families who wanted to donate tissue that might be used in research directed at prevention, diagnosis and treatment of rare genetic conditions. (The desire of patients and families of those with rare genetic diseases to donate tissue was also identified by a spokesperson for the New Zealand Rare Disease Biobank – see discussion later in this report). Academic papers on rare diseases also highlighted problems scientists had when trying to access biological samples from patients and families and the vulnerability of sets of samples when scientists retired or moved to other research centres. (This was also mentioned as a key motivation for the establishment of the New Zealand Rare Disease Biobank). These factors highlighted the need for networking generally and the significance of networks between clinicians, patients and researchers. In these respects, the establishment of EuroBioBank depends on a group of actors that are noticeably under-represented in the UK Biobank initiative – organized support/advocacy groups.

The organisation that assumed the role of promoter and coordinator of the project was EURORDIS – the European Organisation for Rare Diseases.<sup>19</sup> This is an organisation developed to coordinate the activities of patients with rare diseases and their families across Europe. It was founded in 1997, currently receives significant funding from the European Commission, and has grown into an important player in the health sector as a European patient advocacy group – an illustration of what Rose (2007) has referred to a ‘biological citizenship’. It campaigns across a wide range of fields which include improving support services for those with rare diseases as well as strategies to facilitate research on rare diseases. It seeks partnership relationships with corporate sponsors as a way of funding its activities. In these respects, this network of patients’ support and advocacy groups is a key player, not only in networks of patients, clinicians, researchers, service providers, research funders and government officials at a national and an international level, but also networks of international corporates. Other key actors in the European context are EGAN – the European Genetic Alliances Network<sup>20</sup> - a partnership among a set of European groups with a focus on genetics - and EPPOSI – the European Platform for Patients’ Organisations.

In the European context, patients have been significantly involved the establishment of biobanks for rare diseases. They participate in partnerships with corporate sponsors and researchers to develop and manage databases of

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<sup>19</sup> See EURORDIS website <http://www.eurordis.org/>

<sup>20</sup> See EGAN website <http://www.egaweb.org/>

tissues donated by patients and their families for projects that these groups consider useful and appropriate. Against the background of the mushrooming of patients' rights action over the last 30 years, patients and their families, who might be seen as vulnerable in encounters with clinicians requesting tissue donation, are reconfigured as those who have some input into what research will be done, why and how the consent forms are to be designed. They embrace a discursive repertoire of 'empowerment', 'ownership' and 'control' illustrating the ways in which contemporary citizens construct themselves as active biological citizens. According to Nikolas Rose (2007: 147) the active biological citizen is obliged "to live his or her life through acts of calculation and choice" and exert control over his or her own biological destiny – to 'make themselves up' as responsible citizens. That includes the search for information and new knowledge, both personally and collectively.

The Annual EGAN Conference organized in association with the European Human Genetics Conference in Amsterdam in May 2006 had an afternoon session devoted to discussion of joint ventures between patient organisations, science and industry, particularly with respect to the development of databases of human tissue and related information.<sup>21</sup> Key sponsors of the conference were IBM, Novartis, Amgen, Biogen and Genzyme. Just as scientists may position themselves as requiring corporate input to develop their research, so many patient organisations position themselves as unable to deliver to their patient constituencies without corporate sponsorship.<sup>22</sup>

Focus groups, particularly those drawn from community organisations rather than demographic samples of citizens, demonstrate considerable skepticism about corporate interests in the field of genetics (Einsiedel 2003; Du Plessis et al 2004; Petersen 2005). These focus groups often articulate concern about the access by commercial biotech companies to human tissue samples. People in rare diseases advocacy organisations with strong personal interests in genetics research and improvements in diagnosis, prevention and treatment are often less skeptical about commercial companies and more pragmatic about the gains on both sides in any partnership. Rather than seeing themselves as individual 'patients' or the families of patients, they construct themselves as actors in a network with a capacity to influence the actions of clinicians and corporate actors. In the European context they have often positioned themselves as the key advocates of tissues repositories for those with rare genetic diseases. This is relevant for how NZORD positions itself as a key sponsor and advocate for the NZ Rare Disease Biobank.

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<sup>21</sup> Ibid.

<sup>22</sup> EURORDIS includes in the definition of its role applications for grants and activities directed at corporate sponsoring to ensure the continuance of the network  
<http://www.treatnmd.eu/assets/documents/eurobiobank.pdf>

## **Disease specific biobanking initiatives - The case of the Genetics Alliance Biobank**

Patients' rights and advocacy organisations have for some time articulated concern about the factors inhibiting good research on rare diseases. Some of the key stumbling blocks to the development of new knowledge have been the small number of relatively isolated samples in one place and the tendency for researchers not to share the outcome of their study results with others in the struggle to be the first to publish new discoveries and secure commercial rights to certain therapies. The response to some of these problems has been the establishment of patient advocacy biobanking initiatives. Jeffrey Trent of the Translational Genomics Research Institute has stated that initiating a biobank may now have become 'an obligate strategy for patient advocacy groups if you want to make really rapid progress, especially in a rare disease' (Marcus, 2006: 2).<sup>23</sup>

In the USA, the Genetics Alliance Biobank provides a model, not just of patient support advocacy group involvement in the development of a biobank initiative, but of an advocacy owned repository for biological samples, consent forms, clinical and environmental records.<sup>24</sup> This biobank, like EuroBioBank, is a virtual biobank which provides access to information about sets of biological samples associated with rare genetic disorders. The network is controlled by a coalition of advocacy organisations: Angioma Alliance, CFC International, Inflammatory Breast Cancer Research Foundation, Joubert Syndrome Foundation, National Psoriasis Foundation, NBIA Disorders Association and PXE International. The Alliance was founded in October 2003, operates an interactive website and relies primarily on donations.

The Genetic Alliance positions itself as working in partnership with academic and commercial collaborators to achieve better diagnosis of rare diseases and better treatment. Its website acknowledges that scientists have not always acted appropriately with respect to their use of human tissue. They cite the problems of small redundant collections and the lack of good consent protocols. The challenges of research in this field are identified as the small numbers of people from whom tissue can be obtained, the range of different diseases, fragmentation of samples, limited privacy and data security, and poor interactions with donors.

This biobanking initiative takes responsibility for recruiting donors from the advocacy group's own members using strategies directed at increasing trust and ensuring provision of privacy and security. They assert that this strategy for

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<sup>23</sup> Translational genomics involves the 'translation' of scientific knowledge about human genomics into clinical practice. Translational Genomics is a not for profit organisation whose goal is to apply knowledge generated by the Human Genome Project to the diagnosis, prevention and treatment of a range of disorders including cancer, diabetes and neurological disorders like Alzheimer's and Parkinson's disease. See <http://www.tgen.org>

<sup>24</sup> See website for Genetic Alliance BioBank <http://www.biobank.org/>

collecting samples maximizes trust among those who are donors since those who interact with them are those who share an experiential understanding of their situation. This is very different from the challenges of engendering trust in a population-based biobanking system, such as UK Biobank. Dissatisfactions with the way in which consent has been obtained in other contexts inform the consent processes used by the different rare diseases organisations in this alliance of advocacy groups.

The Genetic Alliance BioBank provides a contemporary storage facility and a system for categorizing and archiving DNA, tissue and cell lines. An informatics core is being developed that will encode the identifying information from each donor and store the information in a central data base which is owned by the advocacy organisation. The Genetic Alliance BioBank has let a contract to a private company PreventionGenetics which is collecting and storing samples. Web-based architecture to record and integrate the data is currently being developed. Researchers who want to access samples relating to a particular genetic disorder will submit a standard form to a particular advocacy organisation. The member organisation will then make the coded samples available to the researcher. They are positioned as the go-betweens if researchers want any follow-up samples. This protects patient confidentiality in a context in which breaches to confidentiality can undermine access to insurance and potentially be the basis for discrimination in employment. It also ensures that patients can engage in what is referred to as 'informed decision-making' with respect to how donated tissues are to be used. Member organisations decide what research will be done with the donated samples

The Genetic Alliance BioBank presents itself as a new generation patient advocacy initiative. The discursive repertoire of spokespeople for the BioBank emphasizes patient/family 'management' of the data source, 'empowerment' and 'control'. Patients are recruited by fellow patients or the parents of fellow patients. This constitutes both a redistribution of power and a source of significant power for those who undertake the work of recruiting participants and acting as mediators between patients/their families and researchers. This biobanking initiative is an example of the ways in which biological citizenship (Rose 2007) and biosociality (Rabinow, 1996) is being exercised by patient advocacy groups. As with many new forms of activism, it involves the development of new sets of relations among networked actors, in this case advocacy groups, clinicians, scientists, research institutions and biotech companies. It embraces the model of networks, partnerships and integration of disciplines as well as patient/clinician/scientist collaboration.

The model for the Genetic Alliance BioBank developed out of the actions of PXE International<sup>25</sup> which took the initiative to conduct research on pseudoxanthoma elasticum (PXE) – a rare genetic disorder that affects sight, skin and arteries. The focus was on identifying the relevant genes, patenting that knowledge and

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<sup>25</sup> See <http://www.pxe.org>

developing a diagnostic tool (Terry et al 2007). The initiators of PXE International (parents of children with this disorder) describe their approach to scientific research on this disorder as the creation of a hybrid of academic scientific research, commercial enterprise and patient advocacy activism. They established the PXE International Blood and Tissue Bank and attempted to set up a relationship between scientists and donors which focused on patient/family involvement in decision-making about the use of their donated tissue. As they articulate this relationship: "Research participants are not 'subjects', affected individuals are not 'patients', and the process of becoming involved in the research is not reduced to 'informed consent' but instead involves an informed decision-making process" (Terry et al 2007: 160). Tissue samples from those with PXE and their families are available to all researchers working on projects approved by PXE International.

The advocacy organisation facilitated meetings of scientists with different specialist interests in PXE as well as meetings that brought together those affected by the disease and researchers. As a result of work initiated by the advocacy group, the gene *ABCC6* and the mutations that produce PXE were identified at the University of Hawaii. The discovery of the gene was patented by the founders of PXE and four of the scientists involved in this work. The foundation that runs PXE International was a beneficiary of the patent. In these ways the patient advocacy group has been able to assert rights in the intellectual property produced as a result of their facilitation of tissue and medical record donation. They have asserted their rights to the biocapital arising out of the research they sought to promote. PXE International shares with University of Hawaii the royalties arising out of the patenting of the discovery of the gene for PXE and shares control of the decisions on licensing of the diagnostic test (Rose, 2007: 152).

A genotype-based diagnostic test has been developed and collaborations initiated among the laboratories to which those affected by PXE can donate tissue. PXE supplies pre- and post- genetic counseling and support to those donating tissue and making medical records available. Currently a new treatment for some of the visual problems associated with this disease is being trialed. PXE has played an important part in fund-raising to meet the costs of this trial. A core component of the work of PXE international is initiation of, and support for, collaborations between networks of scientists doing work that is relevant to understanding this disease, including innovation with respect to developing memoranda of understanding between researchers that enhance collaboration rather than competition. PXE currently directs a 19 laboratory international consortium doing research on PXE.

The founders of PXE have asserted the value of support for scientific innovation that is not primarily orientated to commercial gain. The identification of the PXE gene has the potential to stimulate work on hypertension and cardiovascular degeneration because the effects on arteries associated with PXE are relevant



for these conditions. If the discovery of the gene had not involved a patients' advocacy organisation, the costs of research in these fields that was informed by the discovery of the gene for PXE would have been more costly.

On the other hand, the relationship between PXE International and commercial biotech companies is complex. Patrick Terry, one of the founders of PXE International, was recruited as Director of Consumer Advocacy by the biotech company Genomic Health which was set up in the early 21<sup>st</sup> century with seventy million dollars of venture capital (Rose 2007: 152-3). Through these forms of recruitment commercial biotech companies may enhance the commercial value of their products by embracing the ways in which patients and their families constitute themselves as active biological citizens. While patient advocacy organizations in some contexts may broker relationships between biotech companies and other actors, biotech companies may also position biological citizens as actors in networks which are more solidly under their control.

The USA models of patient advocacy biobanks such as the PXE Blood and Tissue Bank depend on the features of a particular national context, particularly on access to money from foundations that dispense large sums of money to charity and a taxation system that provides incentives for such donations. The recent Multiple-Myeloma biobank initiative set up by Kathy Giusti who was diagnosed with multiple myeloma, a rare and incurable cancer of the blood, relied on a significant donation from the Pioneer Fund, a family foundation (Marcus 2006). The money is used to pay assistants at a range of different cancer centres around the country who organize the collection of samples, their storage and distribution to a single collection point at the Mayo Clinic in Arizona. Each time a multiple-myeloma patient has bone-marrow drawn, they are asked whether they want to donate an extra sample to the bank. These extra samples are stored, bar-coded and sent to the central storage site. In this context, it is health professionals rather than support group members who recruit potential donors at a potentially stressful time in their treatment. The medical director of the Mayo Clinic who makes decisions about what happens to the samples; however, these decisions are vetted by the consortium which includes patients.

The forms of patient control over the biospecimen repositories patient advocacy groups are establishing in the USA are heavily dependent on access to large amounts of money from foundations to fund the processes of collection, storage, coding and distribution of samples, let alone the setting up and management of websites. However, the conception that patient advocacy groups might be the initiators of biobanking ventures can and does travel, regardless of context. In the European Union public money rather than charitable foundations funds the activities of coalitions of patient groups active with respect to biobanking initiatives. In the USA, private funding is much more essential. What is emerging is a new role for some advocacy groups as not for profit entrepreneurs in the field of biobanking, the development of diagnostic processes and some treatments for rare disorders.

## **NZ Rare Disease Biobank– getting started**

The establishment of the NZ Rare Disease Biobank is the conception of John Forman, the founder of the NZ Organisation for Rare Disorders and its Executive Director. The initiative to establish such a biobank developed out of John Forman's exploration of information available about the identification and treatment of Alpha Mannosidosis, a genetic disorder which affects the functioning of lysosomes – key ingredients of cells and vital to recycling processes within each cell. This lysosomal disorder generates progressive physical and mental deterioration. John Forman's twin daughter and son were eventually diagnosed with this disorder. It took a long time to arrive at this diagnosis, largely due to its rarity, lack of information about the disorder and the slow pace of its onset. In July 1997, when his children were in their early twenties, John and his wife Judith, a librarian, used the Internet to access information about their children's genetic disorder and establish contact with other parents outside New Zealand. The Internet also enabled contact with a number of clinicians providing treatment for those with the disorder and researchers working in the field. This led to international travel to connect to others with children with rare genetic disorders and ongoing contact via email and websites with parents, patients, researchers and clinicians.

Nikolas Rose has identified the Internet as a key resource for those interested in extending their genetic literacy and constituting themselves as active biological citizens. He highlights the importance of access through the Internet not just to expert discourses about disorders, but also the stories of others with disorders or their family members. Connections to others' narratives about certain disorders, their diagnosis and treatment provide an alternative frame of reference to those of clinicians and scientists and effectively "pluralize biological and medical truth" (Rose, 2007: 142). John Forman's account of his use of the Internet illustrates the significance of this digital technology in establishing cross-national networks of biosociality in the field of genetic rare diseases and the ways in which organizational initiatives in one context can shape developments in other countries.

At a conference on lysosomal diseases in Vienna in March 1999, John Forman encountered delegates from other countries who assumed that he knew about the work of Professor Bob Jolly, a veterinary pathologist at Massey University who had engaged in extensive research on inherited disorders in animals. Bob Jolly had worked on the identification of Mannosidosis, a single gene disorder in Aberdeen Angus cattle in the 1970s. He was the first to identify this rare genetic disorder in a particular herd of cattle and eliminated it by developing a carrier test and culling the carriers from the herd.<sup>26</sup> This work led to the treatment via bone

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<sup>26</sup>Professor Bob Jolly and his associates identified alpha mannosidase, an enzyme that blocks the degradation of glycoprotein which is necessary to inhibit the accumulation of oligosaccharides sugars in cells and maintain the recycling process in individual cells. In this context lysosomes increase in size and impair the functioning of cells. Alpha Mannosidosis is just one of forty-five lysosomal diseases, all related

marrow transplant of people with this lysosomal disorder. It is a classic illustration of the relationship between work among veterinary scientists on rare diseases, particularly rare genetic disorders, and the identification and treatment of rare human genetic disorders. When Forman's connection with the New Zealand veterinary researchers was finally made and the significance of animal models appreciated, it rapidly contributed to the conception of the NZ Rare Disease Biobank.<sup>27</sup>

While there is no systematic repository or register of human tissue from those with rare genetic disorders in New Zealand, there are a number of significant sets of tissues and associated data relating to rare diseases among animals. Most of these databases are dispersed across Crown Research Institutes and universities. The NZ Rare Disease Biobank was conceived as a way of connecting these dispersed tissues samples that would eventually include a human rare diseases data bank. In a context in which the numbers of people with rare genetic diseases is relatively small, the rare disease biobank would, in John Forman's view, "provide added value through attention to both humans and animals". The possibility of a human/animal biobank for rare diseases was also prompted by John Forman's exposure at international conferences to research relating to lysosomal diseases that highlighted what could be learned about human diseases from research on animals. The biobank was conceived as a public good biobank that brought together interesting animal models of genetic disorders as well as human tissue samples.

The proposal for a NZ Rare Disease Biobank was presented for discussion at the national conference of NZORD in 2004, supported in principle by a range of stakeholders (researchers, clinicians, patient advocates) attending the conference, and subsequently incorporated as a charitable company, the New Zealand Institute for Rare Disease Research Ltd. Donations to support this biobank have been used to purchase a freezer for storing samples, for website development and the construction of a comprehensive list of repositories of relevant animal models. AgResearch and the Pathology Department at Otago University are key sponsors as well as the Southern Trust, the Deane Endowment Trust and NZORD. In this respect, this NZ biobanking initiative, like patient advocate biobanks in the USA depends on charitable funding and donations and has established some strong networks with relevant professional groups.

The intention of the directors of NZ Rare Disease Biobank was to develop protocols for human donation of tissue samples in 2006. However, this has been

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to enzyme deficiencies that affect the way sugars and proteins are stored or transformed in cells. About half of these diseases can be treated with bone marrow transplants. Professor Jolly also worked on Batten's Disease (another lysosomal disease) in New Zealand sheep, and has discovered two other important animal models for lysosomal diseases.

<sup>27</sup> See New Zealand Rare Diseases Biobank website <http://www.nzordgroups.org.nz/biobank/>

delayed as a consequence of other demands on the Executive Director of NZORD and that fact that this initiative is occurring against the background of work on the Human Tissues Bill.<sup>28</sup> The Human Tissues Bill focuses on regulating the collection, storage and use of tissues and organs outside treatment settings, including regulation of the collection of tissues and organs for research purposes and from those who are deceased. However, it also addresses a gap in current legislation with respect to the analysis of human tissue (including DNA analysis) taken from living people. The Bill sets out who can give consent for the collection of such tissue, including consent in cases where tissue is taken from children or adults who are not “competent or unwilling to make a decision themselves” (Ministry of Health, 2006). It now seems important to examine the outcomes of consultation processes on the Bill before developing detailed protocols relating to the collection, storage and usage of human tissue donated to New Zealand Rare Disease Biobank by those with rare genetic disorders and their families. However, the intention remains to develop such protocols and eventually create a set of human samples that will complement the sets of animal models for human rare diseases which are currently featured on the NZ Rare Disease Biobank website.

### **NZ Rare Disease Biobank – current developments**

At this stage, the NZ Rare Disease Biobank comprises a networked information source. It has a board of directors that consists of Graeme Milne, a company director, Professor Mike Eccles, Pathology Department, University of Otago and John Forman, Executive Director, New Zealand Organisation for Rare Disorders.<sup>29</sup> In this respect it mirrors, in a formative way, patient-led biobanking initiatives in Europe and the USA.

The key link in the network of databases which make up this rare diseases biobank is the relationship between NZORD and the Pathology Department at Otago University and, in particular, Professor Mike Eccles. There is a possibility that some time in the future, human tissues available for donation to the NZ Rare Disease Biobank will be stored in a freezer acquired for this purpose at University of Otago. Professor Eccles, or someone he designates, will be responsible for managing that repository of tissue and associated information and organising access to it by particular researchers in NZ and internationally, according to access protocols and criteria to be determined by the Biobank board. Access will be overseen by an expert advisory board charged with advising on the merits of research proposals and ensuring compliance with ethics requirements, donor consent and privacy considerations. Given the relative infrequency of the occurrence of genetic disorders in a small population of 4 million people, it is unlikely that those controlling this information resource will be overwhelmed with

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<sup>28</sup> See Human Tissues Bill website <http://www.moh.govt.nz/moh.nsf/indexmh/humantissue-bill>

<sup>29</sup> See NZ Rare Diseases website <http://www.nzrdbiobank.co.nz>

tissue samples. However, given the development of virtual biobanks in other parts of the world, this repository of human tissue could be linked to other data bases.

The NZ Rare Disease Biobank website, which is linked to the NZORD website, includes some basic information about this networked, virtual biobank initiative, including a list of the set of animal models relating to rare diseases that can be accessed at a variety of research sites in Aotearoa New Zealand. The purpose of advertising these animal models is to encourage research on rare diseases utilizing these data. It is anticipated that researchers in other national contexts might be interested in this material and use some of their research money to visit New Zealand and study the animal models available here or directly fund New Zealand researchers to collaborate in projects for which the New Zealand animal model is relevant. It is noted that several projects involving overseas research funders are already under way on these animal models quite independently of the Biobank initiative and this is seen as validation of the scientific and economic benefits of the biobank project. This research using animal models may have significant implications for identification and treatment of human diseases. Increasing the flow of information about sets of human and animal samples is also seen as a way of sparking interest by Masters and PhD students in research in this field. Information about the data sets could lead to the development of thesis projects that used existing research material to generate new knowledge.

The current NZ Biobanking website is due to be expanded so that anyone visiting the site will be able to identify the range of animal models with related tissue samples and then click on a live link which will provide detailed information about the sub biobank, what is stored, where and how the material in the data base can be accessed. Relevant literature on the model or web-links to such literature will be available. It is anticipated that access to this more detailed information will showcase databases available in Aotearoa New Zealand and enhance the possibility of connections among researchers currently working in this field as well as links between New Zealand and overseas researchers. The vision for this website is consistent with the patient advocacy biobanking initiatives that have developed in the USA, particularly the Genetic Alliance Biobank. It is also consistent with the types of procedures for accessing information via the virtual EuroBioBank discussed earlier in this report.

Samples generated for particular research projects may have a limited storage life and access to them may depend significantly on particular researchers in certain institutions. The goal of the NZ Rare Disease Biobank is to ensure that collections that might otherwise cease to be accessible when a particular funding stream ends are known about and potentially available to other researchers. There is some uncertainty about what happens to some of these data sources when researchers retire or move to other research institutions, including those outside New Zealand. New Zealand Rare Disease Biobank is directed at encountering those problems before the culling of non-human tissues data bases

in New Zealand that have been generated to inquire into the significance of genetic determinants for certain conditions. The logging of information about these tissue sets is seen as something that might encourage future research, but also a strategy for ensuring the ongoing availability of some of these sets of samples. This is very similar to the reasons advanced by those active in the establishment of the Genetic Alliance Biobank in the USA.

### **Patient-led biobanking – prospects and issues**

According to the Executive Director of NZORD, most biobanking tends to focus on researchers' interest in getting access to human tissues and the need to ensure that patients exercise informed consent. The assumption is that requests for tissue from people occur in a context of unequal power between professionals and patients and that those prospectively donating tissue need to be protected from scientists whose research agendas (and potentially commercial interests) require access to human tissue. As Forman characterizes it: "the researcher is the one who has the problem, they want to get some tissue." Researchers usually need to convince potential donors about the value of their tissue donation and persuade them to make their samples available and therefore the controls in the form of consent requirements and ethics committee approvals tend to be based on protecting the patient in this power imbalanced relationship. The material available for potential donors on the UK Biobank website is an example of the strategies required to convince potential donors that they should consider participating in this population-based biobanking initiative. Strategies are likely to be different with communities of citizens whose lives have in some ways been touched by genetic disorders. The orientation to biobanking among these groups, especially if recruitment is through advocacy organizations, is much more likely to involve discursive repertoires that assume a joint interest in the availability of tissue for research into specific genetic disorders.

In the New Zealand context, some of those involved in support organisations associated with rare genetic disorders are concerned that there is insufficient research on rare genetic disorders. They recognise that, since these disorders are rare, funding may be restricted for the research of their disorders and the development of treatment options. They are acutely aware that, in the context of competition for research resources, work on the conditions of most importance to them may not be supported by funding sources such as the Health Research Council or the Foundation for Research Science and Technology. In this context they want to encourage research and facilitate work by scientists and clinicians. This sets up a different context for the relationship between researchers and patients and their families. According to John Forman, the families of those with rare disorders are often those who want to volunteer tissue samples. Rather than having to be approached and consider whether they will provide tissue for research purposes, he notes that "it is now the patient and their family that has the problem. They want research on the disease to occur and they need to create

an opportunity for it to occur.” He suggests that family members are likely to say: “I want to donate my tissue and I want my tissue kept” and explore how they can do this.

The desire to donate tissue and contribute to the development of knowledge about particular disorders is consistent with what some social scientists have identified as “hope technologies”. Hope is a core component of the use of medical science to diagnose and treat illness. Technologies of hope extend beyond the specifics of treatment of individuals to organizational strategies that construct hope through actions directed at more knowledge and better treatment in the future for unknown others. Rose (2007: 136) cites Carlos Novas’ work on ‘the political economy of hope’ in the context of biomedicine. This political economy includes many different actors (patients, clinicians, researchers, hospital administrators, researchers and biotech companies) in a complex set of different forms of hope that include relief from suffering, career development, increased profit and altruistic desire for a better life for others now and in the future.

The systematic collection of human tissues associated with the rare diseases biobank initiative has not as yet commenced, although a few tissue samples have already been stored with a view to more formal donation at a later date. People associated with particular support group organisations have made decisions to have tissue samples stored at the time of particular surgical interventions because they were interested in the development of a tissue record relating to those with rare diseases in their families. The decision to make this tissue available for storage and potential use in the future by researchers is driven by the donors’ (or potential donors’) interest in research relating to conditions which have affected them personally, either as patients or family members.

The NZ Rare Disease Biobank is seen by its prime initiator as a way of realigning power relationships among families affected by rare genetic disorders, clinicians and researchers. Patients and families can feel relatively powerless at times as clinicians take samples, analyse results and discuss diagnoses. This arises in part out of inequalities in access to information between families and clinicians. Advocacy groups are directed at addressing that imbalance and constituting those with rare diseases as people with information - knowledge that they access increasingly via the Internet and via their connections to others involved in support groups.

According to the Executive Director of NZORD, those with rare genetic diseases in their family now “know more and we want to be partners in this process.” He suggests that some of the strict ethical controls relating to tissue samples developed in a context where patients and family members were more vulnerable. In this context, ethics committees set up to protect patients and control processes relating to the availability of human tissue for research may be seen by some families as constraining their interests in providing tissue for research purposes. John Forman suggests that some family members with rare

diseases are effectively saying that “we want him or her to have these bits of me, to check this thing out because we want to get on top of this disease – we want it conquered.” This echoes the statements from the Genetics Alliance Biobank spokespeople, particularly those that construct advocacy organisations as partners with clinicians, scientists and commercial organisations. This orientation to the storage and use of tissue samples is also a manifestation of the technologies of hope discussed earlier.

In the future, it will be necessary to set up very robust procedures for the formal donation of tissues by particular individuals or sets of individuals to the NZ Rare Disease Biobank. Regardless of the emotional investment of those who seek to donate tissues and their desire to participate in activities that will facilitate research on rare diseases, it is vital that all tissue donation is subject to a formal consent process. Those involved in the NZ Rare Disease Biobank initiative are clear about the need to establish such protocols and express confidence about a robust understanding of the many ethical, legal and social issues involved, as a result of close monitoring of debates, media articles, literature and proposed legislation, as well as strong personal interest as stakeholders. In a context in which those donating tissue are living lives affected by a genetic condition, it may be even more important that these procedures are detailed and carefully applied. It will be interesting to see how the Genetic Alliance Biobank principle of ‘informed decision-making’ with respect to the use of tissue samples can be implemented in the New Zealand context.

New Zealand health service providers have established some very detailed procedures for consent for the use of samples for research as opposed to diagnosis. Subject to the outcome of consultation on the Human Tissues Bill, particularly the regulation of the collection and use of tissue from living people that includes DNA analysis, the procedures developed by health services for donation of samples for research could provide a good model for those collecting tissue from humans through NZ Biobank. There are also excellent models of consent available in the form of consent templates developed by EuroBioBank. The UK National Cancer Research Institute (NCRI) has also identified a clear and well thought out set of ‘Guiding Principles’ relevant to the management and operation of human biosample repositories. A range of protocols are available that can be adapted for use in the generation and storage of tissues and any associated medical records by NZ Biobank. EuroBioBank and the Genetics Alliance Biobank have also done extensive work on how samples from individuals can be both anonymous and coded to ensure that samples and medical records can be collated and possible contact sustained beyond the period at which tissue is donated. There are also now well developed models for how members of advocacy organizations might liaise with other members of those organizations with respect to possible donation of tissue to patient-led biobanking initiatives.



Since rare genetic diseases in New Zealand include disorders that affect mental functioning and/or communication skills, there may be challenges in implementing informed consent procedures for those with rare genetic conditions. Sometimes family members may be positioned as those who have to make decisions about the use of tissue samples from those with rare diseases. This is a component of the regulation of the use of human tissues which is incorporated into the Human Tissue Bill. Guidelines developed relating to parental consent for the storage and use of sample cards used in newborn screening are also potentially a model for protocols relating to human samples for the NZ Rare Disease Biobank. Parents may be keen to facilitate donation of tissue from their children, sometimes because they hope that tissue donation may generate treatments that would make a difference to their children, sometimes because they are invested in treatments at some stage in the future, even if their children do not benefit.

The NZ Rare Disease Biobank is currently conceived of as a public good biobank where access to material and information is free but subject to donors' consent provisions, ethics approval requirements and satisfaction of an expert advisory group as to the validity of the research proposal and whether the proposed use is a good use of possibly scarce material. Consent forms will need to clearly indicate this to potential donors of tissues to the biobank.

The Executive Director of NZORD is very aware that people donating human tissue are often resistant to its commercialization. At the same time, he has argued that people involved in the rare diseases network are realistic about the extent to which treatment, particularly drug treatment, depends on patenting. He distinguished between the patenting of DNA and the patenting of interventions with respect to diagnosis and treatment that have been developed using non-commercialised human tissue. He has argued that: "in the end you have to get some drug company involved in developing the product if you are to give therapy and they won't invest unless they can put a patent on it and protect their investment".

John Forman was also an advocate of the mutual benefit gained if researchers using tissue donated by people have potential for access to the donors, if prior consent is given by donors for this to occur. He indicated that, among those involved in the rare diseases network, there were potential donors who would not want to be anonymous, but would like to meet the researchers who would be using the donated tissue. In many cases the patient groups and the researchers with particular interests in their disease, often already know each other from attendance at seminars and conferences, or from enquiries made by families after the diagnosis is received. He stated that: "particularly for very rare disorders this is personal investment and engagement... where you don't have this sense that your information is going to go off out there and be used all over the place and you are going to feel powerless as you are in some massive study with a million people". He recalled a recent presentation on the outcome of work on a

genetic mutation associated with a particular condition and the way in which families affected by the disorder in the audience were excited about the possibility that their cell line might have been the source of the new knowledge presented. He argued that in some cases people in the NZORD network were saying: “please take our tissue, please research it.” Many have in the past given material directly to researchers they have met at seminars and conferences and asked them to use it in their research on the disease.

Many patient advocates are aware that material that is not anonymous can be particularly informative about correlation between genotype and phenotype, offering better value to the researcher. There are instances where researchers’ knowledge of the very rare disease, plus knowledge of the particular patients, has led to improvements in patient care, thus giving a significant additional benefit to the patient and their family. In these respects, personal connections between scientists, donors and their families have potential benefits over non-identified access to clinical information and can reinforce the concept of partnership and engagement in the research effort that is valued by many researchers and patients/families.

### **Future directions**

The development of the New Zealand Rare Disease Biobank currently depends on donations and the energy of the Executive Director of NZORD. Progress has been incremental as funds have been limited. To develop into a systematic database that includes useful specimens of human tissue and associated data will involve significant investment. Since the samples are likely to be held in a New Zealand university, processes associated with the acquisition of those samples and their storage will be scrutinized by relevant ethics committees. The consultation processes associated with both the Human Tissues Bill and the public consultation relating to consent, storage and use of newborn blood spot cards will provide resources in relation to the principles to be used in storing and using human tissue for research purposes.<sup>30</sup> In that respect, delay in the development of the human tissue component of NZ Rare Disease Biobank is appropriate rather than problematic. Any move to systematically obtain human tissue samples, store them appropriately and set up procedures for accessing these samples is best informed by current consultations with respect to human tissue.

In the field of New Zealand-based biobanking it is the 40 year old archive of newborn blood spot cards which constitutes the largest biobank. The blood spot cards constitute a significant biological resource (National Screening Unit 2007). However, informed consent procedures for taking blood spots have been relatively informal and no explicit permission has been given for using this set of

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<sup>30</sup> See website for the Human Tissues Bill <http://www.moh.govt.nz/moh.nsf/indexmh/humantissue-bill> and National Screening Unit website for details about each of these processes.

blood specimens for medical research. Testing of newborns is entirely voluntary, but almost 100% of all the children born in Aotearoa New Zealand each year are currently tested. In 2006 the National Testing Centre acquired a new tandem mass spectrometer from the Starship Foundation and is now able to test for a wider range of metabolic conditions, conditions which are included in newborn testing in most other countries in the OECD. The consultation processes currently underway coincides with the availability of a wider range of tests when newborn blood spot cards are screened.

It is legally possible for blood remaining on the blood spot card after the standard range of tests have been run to be used for research that is approved by an ethics committee. However, no research has actually been done using blood spot cards. Parents who do not want their children's cards to be used for any purpose other than metabolic testing can ask to have the samples returned. In practice few parents do this. On rare occasions the NZ Police have used a specific blood spot card to identify a deceased or missing person. This has only been done 15 times in the last 10 years, so use of the cards is a rare occurrence. The consultation document that became public in March 2007 asks readers to consider issues relating to what parents need to know about newborn metabolic screening, the guidelines for consent and refusal, repeat sampling and the storage and use of the blood spot cards. Those submitting responses to the consultation document are asked for their comments on the current practice that any research using the blood spot cards has to have the approval of the National Screening Unit and a NZ Health and Disability Ethics Committee.<sup>31</sup>

Another area in which a new biobank is being developed for research purposes in New Zealand is through the SCOPE project (Screening for Pregnancy Endpoints) based at the School of Population Health at University of Auckland. Associate Professors Robyn North and Lesley McCowan are involved in an international study with seven partners directed at developing a screening test for three late pregnancy conditions – preeclampsia (a high blood pressure condition triggered by pregnancy), fetal growth restriction and spontaneous preterm birth.<sup>32</sup> These are conditions that occur in almost one in five of first pregnancies. The goal is to identify the possibility of these conditions early in the pregnancy and intervene to diminish the chance of them occurring. This will involve identifying molecular markers for late pregnancy complications. At the core of the SCOPE study is the development of a high-quality biobank which would be used to develop predictive tests based on the blood samples of first time pregnant mothers. This biobank initiative has received funding from the Foundation for Research, Science and Technology and the Health Research Council of New Zealand.

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<sup>31</sup> See Newborn metabolic screening programme website: [www.moh.govt.nz/newbornscreening](http://www.moh.govt.nz/newbornscreening) for information about the screening programme and the consultation document on newborn blood spot screening released in March 2007.

<sup>32</sup> See website for the SCOPE project <http://www.scopestudy.net/>

While this research report has focused on setting the New Zealand Rare Disease Biobank initiative in the context of some international biobanking initiatives, the infant and maternal databases associated with newborn screening and research into factors associated with late onset complications in pregnancy are, together with repositories like the Eye Bank, the Tissue Bank and the Brain Bank, more significant repositories of human tissues. These biobanks illustrate the very different sorts of human tissue samples that are being developed around the world, some population-based, like the newborn blood spot tests and some associated with particular disorders and people experiencing particular life cycle changes, such as the women participating in the SCOPE study, or those making their tissues available to the Cancer Society Tissue Bank.

It is unlikely that a major population-based biobanking project, like UK Biobank will be launched in Aotearoa New Zealand in the next ten to twenty years. The costs are prohibitive and the benefits for a small country are very doubtful. A focus on the genetic bases for the diseases of older people such as heart disease, dementia, diabetes, joint problems and various cancers may also undermine attempts to address the socio-economic and lifestyle factors that shape the prevalence and distribution of these diseases – these are currently the focus of public health interventions. At the same time, it will be useful to monitor the development and use for research purposes of a range of different repositories of human tissue, a number of which are currently being used in genomic research or will be subject to genetic analysis over the next ten to twenty years.

Attention needs to be directed at issues that members of the public internationally have identified as fields for attention. They are most crucially issues relating to informed consent, privacy, protection from discrimination on the basis of information about a genetic condition and protection for donors from the commodification of their DNA. Biobanks are costly - they require time to implement good procedures for donors and efficient and systematic storage processes. This means that there will always be significant incentives to make these information resources available at a price to companies. Discussion of how human tissue is used for research purposes and the adequacy of consent processes will need to be a matter of consistent attention. Donors who are part of population-based database collection processes and donors with genetic disorders and their families may well respond differently to issues relating to consent and the use of body tissues. While general guidelines need to be developed, there will also need to be flexibility with respect to the use of human tissue collected under different ethical protocols and for different purposes.

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