AIS SUPPORT GROUP AUSTRALIA
Support for people and families with intersex conditions.
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Submission to the

Australian Law Reform Commission

regarding the

Protection And Use Of Genetic Information.

By

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For and on behalf of the

Androgen Insensitivity Syndrome (AIS) Support Group Australia.

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ABOUT THE AIS SUPPORT GROUP AUSTRALIA (AISSGA) AND THIS SUBMISSION.

The Androgen Insensitivity Syndrome Support Group Australia (AISSGA) is a self-funded peer support group run by volunteers for those with AIS and other intersex conditions, their parents, families and partners. Intersex conditions may be broadly described as conditions able to be medically identified at birth that result in the birth of a child with variations of external or internal reproductive organs or sex chromosomes that are not exclusively male or female. Care should be taken not to confuse intersex conditions with transsexualism. Intersex conditions were previously known as hermaphroditism and are as common as 1 in 1000 births.

The role of the AISSGA is to provide information, direct support, contact with others with the conditions and contact with the medical profession and advocacy services. We have members Australia wide and overseas. Our membership includes medical practitioners considered by the AISSGA to provide best practice treatment of members, particularly those practitioners that challenge older inappropriate practices by advocating full disclosure to patients about their condition and challenging early surgical based intervention. Our membership also includes genetic counsellors and geneticists. We have strong links with the Genetic Support Network of Victoria, the Royal Children's Hospitals in Melbourne and Brisbane, the Mater Children's Hospital in Brisbane and various other paediatric hospitals around Australia.

The AISSGA has a direct interest in the outcome of discussion, formulation and any guidelines regarding the use, or ultimate statute protection of genetic information. Those with intersex conditions have the same ultimate desire as most others in the general population, that of being able to live without fear of discrimination or identification on the basis of their medical condition.

This submission has been prepared to outline the points of view of members of the AISSGA about various issues raised by the ALRC inquiry into safeguarding the use of genetic information. The AISSGA submission considers issues raised in the ALRC issues paper as well as others of concern to members.

ABOUT THE AUTHORS.

Andie Hider is the current second term Secretary and Medical Liaison officer of the AISSGA. She has formerly held positions as the State representative of the AISSGA for a number of States. Andie represents the AISSGA on medical projects and is the principal AISSGA point of contact for the medical profession. Andie is a tertiary qualified mechanical engineer with a defence science related research background and she also has completed post-graduate subjects in science and law. She is also an adviser on Equity and Diversity issues, Gay and Lesbian Liaison Officer and harassment contact officer for her employer. Andie is a member of the Australia and New Zealand Forensic Science Society and regularly reviews medical and scientific journals, with a particular focus on articles related to medical and scientific ethical issues.

Tony Briffa is the current second term President of the AISSGA. He has previously held other positions on the support group's committee, including that of Secretary. Tony represents those with intersex conditions on Government and other advisory committees. He was instrumental in successful inclusion into Victorian Equal Opportunity legislation of protection for people with intersex conditions to live as their self identified gender. Specific provision for protection of people with intersex conditions in this way is a world first. Tony has wide experience as an advocate for people with intersex conditions, being quoted in many magazine and newspaper articles about the subject and appearing on television. Tony is an engineer with considerable experience in the aviation industry and with a broad technical background. He regularly reviews medical journals to keep up to date with current medical and scientific treatment practices. Tony is also a foster parent.

GENERAL

The overarching AISSGA position on the use of genetic information is that it should only be used for a genuine medical purpose with express consent of the provider of the human tissue sample to which the information relates. We believe that use of genetic information for what we regard as a solely commercial purpose to be contrary to the benefit of society as a whole. Our definitions of genuine medical purpose and solely commercial purpose are explained in detail later in this submission along with our reasons for using the legal standard "reasonable and prudent person" test to determine into which category the use of genetic information would fall.

The position of the AISSGA is partly based on a continuing presumption of the right to privacy (notwithstanding decisions regarding the common law in this respect) and partly on foreseeable outcomes of the continuing advancement of medical and allied sciences related to the field of genetics. Whilst the effect of legal decisions such as the "possum abattoirs" case on the right to privacy will no doubt be the subject of much discussion amongst legal practitioners, consideration of issues like the potential effect of foreseeable advances in genetic science and the potential use of information derived from those advances should be subject to widest possible consultation of consumer groups. As with other fields of science, genetic information should be used in an ethical, responsible way that serves to benefit society as a whole without being detrimental to any specific minority groups.

Although not an issue subject to investigation in the ALRC inquiry as such, it is important to keep in mind in any discussion of issues surrounding the use of genetic material, that genetics is an area where we are certainly more than the sum of our parts.

WHO SHOULD BE ASKED ABOUT PROTECTION OF GENETIC INFORMATION?

The question of who should or should not be considered a consumer group of genetic information is, of itself, probably a subject worthy of considerable discussion. The position of the AISSGA is that the broadest possible meaning should be given to the term consumer groups, indeed use of genetic information is something that has the potential to impact on every person and every generation in society, therefore the safeguarding of the use of such information is in the interest of all groups in society rather than a select few. It follows that widest possible consultation of community groups will allow formulation of safeguards that best represent the current and foreseeable future concerns about the use of genetic information.

WHAT IS THE AISSGA VIEW OF WHAT CONSTITUTES GENETIC INFORMATION?

Society, by the very nature of it's diversity, provides not only a tremendous "genetic pool" from which to gather information, but many different views of what constitutes genetic information and potential uses for that information. If there is a genetic version of the "golden thread", it must surely be that genetic information is all about all of us and belongs to all of us. It is often seen as being essentially medical or scientific in both origin and application, many will not appreciate that genetic information is about the origin and future of us all.

The medical explanation of AIS and other intersex conditions involves conceptual understanding of both cellular and genetic science. Most with AIS will have had their condition explained to them in a way which includes the genetic origin of the condition. A diagnosis of AIS can often be confirmed at a genetic level. Members of the AISSGA have recently been invited to take part in a genetic study which purpose it is to understand the genetic origin of organ formation. Intersex conditions are of particular value in this context as outcomes of genetic variation are directly measurable as the formation of testes, ovaries or dysgenic tissue[1]. Many members of the AISSGA thus have a good understanding of genetic information and it's uses.

Genetic information will generally be seen as medical information by our members. It follows, that if genetic information can be used to confirm existence of a condition or pre-disposition to a condition, that ultimately all genetic conditions will be able to be traced back to their genetic origin. In our opinion herein lies the first cautionary proposition: information held in medical files that has not been directly derived from genetic testing may ultimately one day be able to be linked to a genetic origin and therefore indicate an underlying genetic precursor, so all personal medical information should therefore be regarded as genetic information and safeguarded accordingly. Our members would agree with both the underlying proposition and the subsequent application of safeguards to all medical personal medical records, with deviation being the rare exception.

VARIATION OR DEFECT? AND WHO GETS TO DECIDE?

Ask a group of people to draw the line between a genetic variation and a genetic defect and you will elicit as many different points of view as there are people in the group. Like all such things, the line drawn will depend on their life experience. For those who live with a genetic condition, the line between a variation and a defect is likely to be very different to that drawn by someone not obviously affected.

The medical view of the difference between a variation and a defect is yet another issue. The medical profession have historically held the view that any variation from their perception of normal development should be considered a defect. "On the other hand, scientists still tend to treat as abnormal anything that doesn't fit the male-female model."[2] Whilst this is no longer universally the case, many are looking to genetic variations to provide clues about the development of all of us, dissenting views would seem to be mostly held amongst those involved in the field of medical research. Indeed, it might still be considered an accurate proposition that the view of genetic variations may be divided into the views of medical practitioners and the views of researchers.

Members of the AISSGA and other intersex groups worldwide would, in the vast majority, consider their conditions to be a natural variation of development, rather than a defect. "Above all, I would just like to reiterate what I say to people when discussing intersex conditions: Intersex is a variation, not a defect"[3]. Language and societal perception play a large part in the way any person with any naturally occurring biological variation see themselves. Societal perception will be influenced by such things as education, personal experiences, perceptions of the medical and scientific communities and the availability of such things as peer support and counselling. A recent report regarding termination of pregnancies after disclosure of existing genetic conditions[4], was critical of the way in which such information was delivered. The report adversely commented about the number of unnecessary terminations of children with genetic conditions that did not impact on quality of life in any significant way. It was, significantly, the way in which parents were given the results of genetic tests without accurate information or access to counselling that influenced the decision to terminate the pregnancy. Genetic information is highly persuasive when decisions about termination or continuation of pregnancies are made.

The AISSGA was recently contacted by a member who spoke with parents considering termination of a pregnancy, because of a positive genetic test for AIS. They had received no more information about the condition than it was considered a major defect and had been offered no counselling or support by specialists. Their GP, who was also the GP of our support group member, was concerned about their decision to terminate a child who would have no serious health issues simply because she would be born female but with a male chromosome pattern. After speaking with the GP the parents were put in touch with our support group member who invited them to meet her at her place of work. What the parents discovered was the woman they met was an attractive, intelligent, senior manager in a large corporation. They realised the little information they had been given by specialists was largely misleading and based on a medical perception of the life of someone with a genetic "defect" rather than any real contact with those with AIS. The parents understood that termination of the pregnancy was clearly inappropriate. Incidents like these are commonly encountered by members of many support groups for those with genetic conditions.

Other concerns regard the underlying medical reasons for genetic variations of development. As more is understood about human development, and more information regarding development is able to be identified from scientific investigation, the more the scientific community realises the valuable role those with genetic variations play in understanding our origins. Mother nature has historically been very unforgiving of those who would presume to know better than her the reasons for various biological processes taking place. Using any genetic information in a way that is scientifically irresponsible has the potential to cause great harm to us all, especially by eliminating something from the gene pool that is not fully understood.

IS THE PUBLIC PERCEPTION OF USES OF GENETIC INFORMATION REFLECTED IN REALITY?

Ethical use of genetic information has largely become a public debate and media interest in this area of science and medicine has been substantial. Hope for many that suffer from life threatening or debilitating conditions comes in the form of regular announcements of medical and scientific breakthroughs. Often these announcements are of discoveries of the gene said to cause a particular condition. This gives the impression to many people, if not the majority, that genetic information holds all the answers to their most asked medical questions. Perhaps more worrying is the belief amongst many that genes work in isolation, each responsible for a single task or outcome. This has led to many misunderstandings about genetic information and the way in which it may be used, especially the belief that genetic pre-disposition to a condition means inevitable physical manifestation.

Geneticists will quite rightly point out that genetic information, whilst of considerable scientific and medical value, is not the ultimate medical solution that some believe it to be. Genetic information is a starting point, not the final solution to all medical ills. It is a good place to begin to fill in some of the gaps that exist in our understanding of the human body. The ability to determine the way organs form for example, perhaps leading to an understanding of why some may fail and the eventual discovery of ways of preventing such things as kidney failure.

It is doubtful if many of the non-medical institutions seeking access to genetic information truly understand the nature or use of that information. It is likely that public belief about and media portrayal of the practical application of genetic information has led to the belief that genetic testing will provide information of direct use. At best, such genetic testing is likely to provide indirectly useful information, at worst, lead to discrimination of the worst type. What is clear is that education of many groups about the uses of genetic information is necessary and should be seen as a vital part of not only the community consultation process, but also to create an environment where informed consent is possible. It is important that such education should leave no doubt about the limitations of genetic information and the dangers of assumptions as to the inevitability of outcomes associated with positive genetic tests.

WHO REALLY "OWNS" GENETIC INFORMATION?

Intellectual property rights is an area of law that has advanced considerably in the last ten years as a direct response to the recognised world wide economic importance of advances in technology. Applications for patents involving medical research have increased dramatically in recent years. Genetic research has not escaped this trend. Intellectual property is generally categorised as having had some development or furtherance of an idea before it is accorded protection under intellectual property law although this is not a definitive rule by any means.

It is generally understood that there is about one tenth of one percent difference in the genetic make-up of each of us. By any standard this is a very small percentage. It is nonetheless very important. It is the interaction of that point one percent genetic difference with all of the other "commonly" shared genes that makes us each genetically unique. Often, it will be that point one of a percent that researchers are interested in. As an example, the action of Teleomeres in the process of cell reproduction is now believed to limit the number of times a cell may reproduce, hence being directly responsible for the ageing process. Studies of children with a rare genetic condition that results in premature ageing, led to the conclusion that cells don't just wear out, there must be something that controls how many times they can reproduce. Understanding the action of Teleomeres has led to possible therapies for this condition, with the flow on effect that it may be possible to increase not only the life expectancy of these children, but the life expectancy of all of us.

To whom does genetic information really belong then? In the above example and the earlier example of the role of intersex conditions in understanding organ formation, it is clear that the point one of one percent is of considerable value as a scientific tool. Such research is also clearly of benefit to all of society, not just those who provided the genetic information for the research. Should information about the roles of particular genes be considered intellectual property? Most would argue it does not fit the description of being developed or having some act done in furtherance that advances the information, the basic biological information exists in undocumented form. It could be quite rightly argued that the processes used to discover the action of the gene is intellectual property, or that any process used to treat conditions identified as a result of the genetic information discovered is intellectual property. It follows, that if the researcher does not have intellectual property rights over unique genetic information because they have neither developed it nor done some act in furtherance to advance it, then neither does the person from whom the genetic sample was obtained.

There are, however, other physical property rights to be considered that apply to the person from whom the sample came. Rights to physical integrity originate from common law. Statute protection is afforded against trespass upon a person in many forms. Medical procedures carried out without adequate consent have been held to be an assault at law. Recent case law has established property rights in relation to tissue samples and there has been criticism of various institutions obtaining and cataloguing samples from deceased persons without permission. Clearly, legal recognition of ownership of human tissue samples by the person from who they were obtained is becoming widely accepted.

It is clear that little research would be undertaken were there no likelihood of protection of ownership for the information involved. For this reason, any developments resulting from information gained from genetic samples or acts done in furtherance, such as developing "gene therapy" processes using synthetic

genes, should be considered intellectual property and treated as such. Basic information though, such as the mapping of the human genome and identification of the role of particular genes in human development, should belong to everybody. Tissue samples provided for research, should always remain the property of the person from whom they came, subject to consent for research provided expressly by their owner.

SHOULD GENETIC INFORMATION BE AFFORDED SPECIFIC LEGAL PROTECTION?

The current level of access to genetic information is a direct product of continued advances of medical science and technology. The genetic information being discovered as a result of research seems to be growing at an exponential rate with new discoveries being announced almost without fail every week. Genetic testing is potentially the most invasive, personal and intrusive form of medical investigation, both for the extent of information it may reveal and the accuracy of the results. The information gained can be accessed and analysed repeatedly once it has been recorded.

In order that safeguarding of genetic information keeps pace with the rapid advances of medical science, we believe specific legislation is needed that relates solely to protection of genetic information. Legislation that currently protects medical information is of questionable value, given the increasing number of non-medical groups attempting to gain access to it. To treat genetic information in the same manner as other medical information, given the information potentially revealed by genetic testing, is to afford nowhere near the level of protection it deserves. Reasons for the AISSGA position on this issue are discussed in detail later in this submission.

A POTENTIAL TEST TO CLASSIFY THE USE OF GENETIC INFORMATION?

Central to any discussion about the use of genetic information, is going to be the question of what constitutes a genuine reason for the use of such information? As briefly outlined earlier in this submission the position of the AISSGA is that, with the exception of closely regulated use for law enforcement, use of genetic information should only be for a genuine medical purpose without causing or indirectly invoking any detriment to any community group. The question we would apply to test if a genuine medical purpose exists is; does some demonstrable or foreseeable medical benefit exist for a specific patient or group of patients? If so, then it would classify as a use for a genuine medical purpose. Conversely a solely commercial purpose would be a use of genetic information that does not benefit any patient or group of patients in that way. Use of the term "patient" is a deliberate departure from the term "persons" as "medical benefit" is a deliberate departure from "benefit" as both add a level of specificity that we feel should be part of the test. Both employer and insurance groups would fail the above test for reasons that their relationship with those they would seek to test is not as patients or there is no medical benefit.

Few would argue that commercial medical concerns, such as pharmaceutical companies, have a genuine interest in having access to genetic information. As there is a demonstrable benefit to patients as a result of their research they would pass the test described above. Even though the use would generate commercial benefit, there is still the ultimate benefit to patients that derives from the research. Clearly, research institutions that form part of public instrumentalities, such as universities or medical centres, would have access to genetic information under the test we describe above even if their research is privately funded.

A test of this type could be administered by the legal standard "reasonable and prudent person" as a full understanding of genetic science is not required to consider an application of use in the context of the above principles, it is merely a test to determine in which category the application of use falls. Subsequent questions of ethical research matter and the way genetic information is used for that research may be decided by a Human Research Ethics Committee made up of appropriate members.

WHO SHOULD DECIDE WHAT IS AN ETHICAL USE OF GENETIC INFORMATION?

Human Research Ethics Committees (HREC) have a place in helping to safeguard the use of biological samples for genetic research, but do their ethical points of view represent those of society or of consumers of genetic information? Are societal views, the context in which applications for genetic research should be considered?

Genetic information is about all of us and for all of us. Medicine and science have become allied in a way never imagined 100 years ago and both have become self sufficient in a way never imagined 100 years ago. Both medicine and science have an origin that is beyond self service though, that of serving society. It follows that the fields of medical and scientific research should be guided by societal views of ethical behaviour. Where this proposition fails, however, is where there is not sufficient societal understanding of the work undertaken by researchers; research using genetic information is one such area where a lack of general understanding exists. In our view it is simply not sufficient to entrust decisions about the use of genetic information to HREC simply because there is a lack of societal understanding about this field of research.

Generally speaking, good research subject to peer review will withstand scientific scrutiny, but the ethical considerations about the use of information gained from research should be subject to societal review if the interests of society are truly to be served. HREC, by their very nature and limitation, also consider use of genetic information in a fairly narrow context. For genetic information to be used in a way that society truly considers appropriate, it is important to have in place a system that allows for consideration of use of genetic information in the context of expression of contemporary societal concerns.

There are various Government Standing Advisory Committees (SAC) that consider many issues in light of current values, both domestic and international. Establishing and appointing positions on such committees is often a difficult process, but the difficulty encountered is more often a product of the gravity of the issues for consideration than any administrative complexities. Protection and consideration

of use of genetic information is something about which community, consumer, industry, medical, scientific and Government groups all have varying opinions. Ensuring the interests of all are served appropriately means ensuring all are heard in an effective way, establishing a genetic SAC would be the most appropriate approach. For such a committee to be both seen to be and to operate in reality in an way that is representative, there is a need to ensure sufficient representation of all groups that have an interest. It is particularly important, as the largest consumer group of genetic information, that community groups are given at least equal representation as other groups, such as industry, science and medicine.

A genetic SAC is going to see interrelationship of opinion vary considerably depending on the use of genetic information being considered, but it is important that such variation of expression be considered as it is an indicator of societal interest in the outcomes of decisions. As an example, church groups and many of those with genetic conditions might oppose genetic pre-natal screening for the purpose of terminations of pregnancies. Each group would potentially have their own reasons for opposing the use of genetic information in this way, but it is important that each is heard and gauged in the context of contemporary societal values so that the interests of all are considered appropriately.

A committee with such wide representation is also more likely to offer and consider alternative ways of effectively dealing with demands for access to genetic information. A difficult role for any group considering the use of genetic information, is the "Pandora's box" principle. Research involving genetic information is very rarely going to have an exclusive end use and herein lies the danger of irresponsible control of the use of genetic information. Information is one of the most difficult things to safeguard adequately, witness the many millions of dollars spent by industry annually in Australia alone in an attempt to safeguard industrial information. As an example of how serious a threat to world economies theft of industrial commercial information is now considered, the United States Federal Bureau of Investigation has a direct responsibility for investigating such thefts under the provisions of espionage legislation. Of all the forms of information available, surely genetic information has the potential to be the most damaging to humanity if misused.

Legislating against the misuse of genetic information has already taken place in certain circumstances. There will always be debate about the appropriateness of such action. In the end though, Government is intended to be representational of the will of the wider community and, in such a role, has a responsibility to ensure that the use of genetic information for such things as pre-natal screening, represents a use that is desired by an appropriately educated greater community. A genetic SAC would also be responsible for considering potential avenues of education of the wider community about the uses of genetic information.

A genetic SAC is never going to replace the need for the institutional HREC, but would provide a more thorough and broad ranging test of societal opinion than is currently considered by individual institutional HREC. Provision of what are considered to be the minimum ethical standards required for consideration by HREC would be something that a genetic SAC is able to provide in the context of better understanding of societal views. As a representational group for consideration of such things as legislative changes, a genetic SAC would be invaluable in ensuring public interest in Government decision-making is represented.

A POUND OF FLESH, RESEARCH AND GENETIC DATABASES.

Medical and scientific research has the potential to directly affect quality of life in a way no other issue is able for every one of the billions of people alive today and for future generations. Care needs to be taken to ensure that such research generates outcomes that benefit society whilst allowing for responsible use of information obtained as a result. Medical and scientific ethics are becoming specialist fields in their own right, as evidenced by those specifically focussing their careers on this aspect of research. Striking a balance between encouraging scientific and medical research and protecting the interests of those such research may ultimately serve, is truly walking the tightrope.

Genetic databases provide a primary source of information for genetic research. Many tissue banks and the genetic information obtained from them have taken considerable time to develop to the extent required for them to be useful research tools. The collection, collation and maintenance of such material has not been without controversy and increased use of human tissue samples kept in this manner is likely to generate further question as to the history of the material.

Central to any discussion about research using human tissue samples are two issues; the way in which such material is obtained and the way in which consent for initial or subsequent use is obtained, especially for research. Other concerns, such as privacy of information relating to obtained samples and ethical use of information obtained as a result of research, are obviously not issues if there is no material to conduct research in the first place.

Much of the controversy surrounding the use of human tissue samples, has been because of those the AISSGA would term "grey samples". This is material that has been obtained ethically and lawfully for one purpose, but is then used for a purpose not expressly consented to. Examples of "grey samples" would include those later used for research but initially obtained for patient tests, as a result of surgical excision and samples obtained as a result of general infant screening tests. Access to human tissue samples for research would become restricted to an unworkable extent were it necessary to obtain express consent from the person supplying the sample each and every time some new research project were approved by HREC. There is still a need, however, to ensure that adequate consent is provided prior to obtaining samples in the first instance. As a minimum standard, any person that provides a sample for any specific purpose or purposes, should have explained the potential for the sample to be later used for research before consent is obtained. We believe that it is ethically indefensible, especially in the contemporary ethical environment, to obtain samples without a person having explained to them at least in broad terms all purposes for which that information may be used.

Consent to the taking and use of human tissue samples raises a number of questions in and of itself. Firstly there is the question of informed consent. Can a person who has no real understanding of the potential ultimate uses of genetic information, truly give informed consent for the taking of a tissue sample for research purposes? Should express consent be obtained each and every time a researcher seeks to use a tissue sample for research? If consent is obtained in an environment where it is felt that adequate understanding of the nature of research exists, is such consent perpetual? Perspectives of appropriate research are dependent upon personal experiences and therefore highly subjective, can a person who has consented for use for a specific purpose or purposes, withdraw that consent if their view later changes?

The majority of researchers seeking to have access to genetic information undoubtedly do so for reasons that they genuinely believe to be altruistic, however, their views like the views of all others in society are influenced by their own experience base. It is very likely that issues of necessity or benefit will continue to arise when researchers apply for access to tissue samples for research. But where the interests of medical and scientific researchers are perceived to be in competition with societal standards, is this truly the case or are societal standards, as discussed earlier, built upon a foundation of generalised beliefs rather than actual information?

Perceptions based on life experience are very difficult to quantify because of their subjective nature. HREC vet most applications for access to human tissue samples as a moral and ethical "yardstick" against which numerous competing interests are measured. Currently, HREC effectively act as a "snapshot" of societal structure, purporting to represent social ethical concerns applied in the context of scientific, medical or ethical expertise. Although each of those persons making up an HREC are not immune to their own subjective views, like any committee with varied representation, these are kept in balance by the views of other representatives. In context of current frameworks, could an individual's consent to use of their tissue sample, override consideration by an HREC? The answer is that this is unlikely because of the limited understanding of the nature of the ultimate use of the sample by the person consenting to its provision. This brings us back to consideration of the earlier questions about express and informed consent and the effectiveness of such consent.

Wider understanding of issues surrounding the protection and use of genetic information are not what could be sufficiently described as general public knowledge. The complexities of the issues involved require at least a reasonable base level of specific medical or scientific knowledge. To ask a person to give consent to the use of their tissue samples for research purposes, without this base level of knowledge existing, challenges the very concept of informed consent. It is clear that any person who refuses consent to the taking of a personal sample to be used for research, has every right to refuse. It could be argued that some may subsequently give consent if their base level of knowledge about genetic information and research is improved to the extent they have sufficient understanding of the issues involved. Counter to this argument, would be concerns about coercive application of provision of information to the subject that does, or tends to, dismiss without adequate basis any concerns they may express. It follows that in the contemporary HREC operational environment, any individual consent given for use of human tissue samples should be considered in the light of the general level of societal understanding of the nature and use for such information, not the level of understanding possessed by panel members of the HREC. Construction and selection of panel members of HREC, when the above is considered, becomes especially important.

It has been well established at law, that consent may be withdrawn at any time. There are a limited number of circumstances in which actions may be carried out despite withdrawal of consent, but generally these are the exception rather than the rule. The question of continued consent and at what time that consent may be considered to be withdrawn either verbally, in writing, or by action, has also been the subject of much legal consideration. How then, if at all, does consent for use of human tissues samples differ from consent for other medical interventions or, for that matter, any form of consent. Legally and ethically speaking, the likely answer is that consent given for provision of a human tissue sample for research does not differ from any other form of consent. This being the case, it is important to have

mechanisms in place to not only first record consent to use of a human tissue sample, but something that details the degree to which a person understands that use and, ultimately, allows for withdrawal of consent at any time. Human tissue samples for which consent for research use has been withdrawn, **should be destroyed, not de-identified.** Whilst removing such details as name, address, date of birth, patient UR or file number etc appears on the surface to de-identify the sample, in reality this has not happened. It is likely that a human tissue sample given expressly for research would be sufficient to be fully profiled, it is therefore possible to attribute that "de-identified" sample to a family line or single person. This is an especially important point, as genetic information like no other has the potential to be used to profile pedigrees of more than one member of a family. Consent by one family member means that information about the rest of that family may be elicited from a single human tissue sample. This should be clearly explained to any person from whom consent to provide human tissue samples is sought. We therefore believe that all possible steps should be taken to enshrine consent relating to human tissue samples in a way that the law demands for consent as a general principle. Where informed consent is questioned, the onus should be upon those using the information to demonstrate that informed consent was granted and was current at the time the dispute took place.

In addition to legal and ethical issues that must be considered for samples provided with appropriate consent, are those "grey samples" previously obtained without such express consent. It was once common practice to retain human tissue obtained for a variety of reasons, but contemporary application and understanding of ethics allied to recent controversy and media attention focussed on human tissue samples taken and used without permission, would suggest that societal views of this practice are not favourable. Understandably, there are ethical and moral concerns about the use of human tissues samples obtained and used for research without express permission. Recent cases that examine the issue of "ownership" of tissue samples, challenge the view that excised tissues had no owner and were therefore able to be used at the discretion of medical researchers. It is becoming increasingly obvious that even though intellectual property rights over genetic information are questionable, property rights remain with the person from whom the sample came, legally, ethically and morally. Do these shifts in ethical and legal viewpoints operate retrospectively? Whilst "grey samples" were obtained in an era when such issues had not been examined, it is undoubtedly their use when having due regard for current ethical and moral standards that must be tested. By any current standards, the use of human tissue samples obtained without express permission must surely fail any reasonable ethical and moral test.

There is a view that the medical and scientific research communities are putting the cart well before the horse in even having such tissue banks as exist currently. Many hold the view that there is insufficient understanding of genetic information and genetic conditions generally, and that there are insufficient safeguards in place to protect genetic information. Liaison with established support groups rarely takes place and, as such groups are often made up of those living with genetic conditions, the perception is that research is geared towards elimination of medical "defects" in the distant future, rather than addressing quality of life issues in the here and now. It is often the case that those with genetic conditions do not see themselves as being sufficiently different to mainstream society, or even apart from mainstream society, to warrant being genetically eliminated. Society depends upon the right for diverse groups to exist for many quality of life issues, the right for future generations to exist when they do not believe their condition serious enough to necessitate medical elimination should be guaranteed. It should not be up to medical or scientific communities to become genetic societal constructionists, rather they should seek wider consultation with community groups to ensure the work they do is directed appropriately toward

improving quality of life for every group in society. Inclusion of representatives from genetic support groups and other consumer groups on HREC should be seen as an important step towards being socially and ethically responsible and responsive.

Like many forms of information, once available, genetic information will be subject to future use for many purposes never envisaged by those conducting the original research. Whilst forecasting trends is difficult in many areas of science, interest in genetic information from a variety of groups suggests that advances in testing and recording technology will be keenly followed. Furthermore, because human tissue samples and the resulting genetic information are able to be stored for a period of time that is to all effects indefinite, technology and science have more than ample time to "catch up" with the information stored waiting to be discovered. HREC and a Government SAC, should monitor research and on-going use of genetic information using a well established reporting framework. Use of any information that results from research using human tissue samples, should also be carefully regulated. Lack of understanding of genetic information and genetic conditions, has already seen uses of genetic information that are considered socially and ethically irresponsible by some groups. Lack of tolerance allied to misunderstanding of information is a socially dangerous combination, it is all the more disturbing when associated with the medical profession. Scientific protection of personal genetic information is often adequately controlled by current safeguards, it is the post research application of that information that is often subject to question.

It is inevitable that current commercialisation of scientific research will dictate to some extent the way in which safeguarding of genetic information is able to take place. Whilst commercial research bodies still provide substantial benefit to society, care should be taken to ensure they are regulated appropriately as with any other industry. Commercialisation rights of research based on genetic information should be denied where breaches of ethical safeguards occur.

The underlying current of all discussion about research is that medical and scientific ethics should be primarily influenced by societal needs. Arguments that most members of society do not understand the nature and uses of genetic information do not carry sufficient weight to excuse past inappropriate action, such as the collection of "grey samples", nor justify current practices of ethical concern, such as the use of "grey samples" for current research. Steps should be taken to allow for wide public education and discussion about genetic information and adequate controls of scientific use, before further applications are made to obtain human tissue samples and use them for research. Researchers must be included as part of society when discussions about ownership take place, but whilst their work is very important, they still represent a minority when considered against humanity as a whole. It is very important to keep this perspective when looking at the nature and needs of research in the context of whole of society interest.

In a perfect world, it would be possible to conduct research using genetic information without introducing the human element such research commands thus taking a pound of flesh without taking a drop of blood. Unfortunately for researchers, like the literary example, conducting research is and must be inevitably tied to greater human concerns.

PRIVACY

The principle of the right to privacy was one of the first to be enshrined in common law and statute. The right to privacy has frequently been tested in courts overseas and here in Australia and there are many circumstances in which the right to privacy is balanced against competing interests. The fact that statute exists that considers privacy to be resident in the first instance when examining any need to breach it, is a clear indication of the level of presumption attached to the principle.

Medical information has the potential to be very damaging to a person. In addition to concerns of unauthorised release to third parties, is the need to ensure that disclosure to a person to which such information relates is done in a way that gives appropriate consideration to the sensitivity of the situation. This is particularly important where such information is of a genetic nature and may directly affect not only the person tested, but also current and future members of their immediate family. Feelings of guilt associated with passing on a genetic condition to a child can be very complex and require specialised assistance to understand and cope with, disclosure without appropriate support is simply not acceptable.

Disclosure of the results of genetic testing has become very specialised. Not only have genetic specialists become increasingly aware of the necessity to provide carefully considered guidance in such circumstances, genetic counsellors are becoming indispensable now that genetic testing is more frequent. Genetic counselling is becoming a more specialised profession in and of itself, with some genetic counsellors choosing to develop expertise exclusive to a particular group of conditions.

Accidental or unauthorised disclosure of medical information is not unknown. The AISSGA is aware of incidents of this type amongst our membership. As an example, one woman with AIS (who was not aware of her condition) was informed of having her condition by her bank upon application for a loan. She had signed a release form allowing the bank access to medical information for the purposes of the loan. At a subsequent interview regarding the loan application, she was asked what is Androgen Insensitivity Syndrome? Disclosure of information of such sensitivity in circumstances like these is clearly inappropriate. Why does an employer need to know information of this type? AIS has no limiting effect on employment prospects, it would simply be one more record of information that should remain the sole interest of the person with the condition and their chosen medical practitioners. It follows that the more people that have access to any particular piece of information, the greater the chance the information will be passed on to someone else. It need not be as a result of malicious intention that information is released inappropriately, but such things as accident or simple misunderstanding. Limiting access to genetic information is the only guaranteed way of minimising the chance of unwanted disclosure.

Currently, there are certain circumstances that attract a moral and ethical responsibility to disclose genetic information. Whilst there may be certain medical practitioners who are reluctant to disclose information because of diagnosis based patient concerns, it is generally accepted that honest and full disclosure to a patient is ultimately in their best interest. Can it be said of insurer and employer groups seeking access to genetic testing, that they are morally and ethically obligated to act in the same manner as the medical profession? Differing reasons for employer and insurance groups applying for access to genetic information and the way in which that information is subsequently treated, provide one of the most compelling reasons for limiting access to genetic information. It would seem an unconscionable state of

affairs that a prospective employer or insurer knows more about an applicant's genetic status than the applicant themselves and it is clearly unreasonable for such groups to disclose genetic information to the applicant or to expect this of them. It is better that genetic information is excluded from access by employers and insurers, than to try to safeguard or regulate use once the information has been obtained. Moral and ethical concerns about recording and disclosure of genetic information are better left in the hands of those who specialise in such fields of medicine.

The contemporaneousness of legal safeguards put in place to protect privacy is questionable. Like all matters of law, statute provisions are generally only tested when some question as to application arises with amendments subsequently made where appropriate. This is hardly surprising as it is unlikely that those who originally sought to have privacy protected as a common law principle understood the tests such principles would face as the nature and needs of the law changed. No reasonable person is likely to challenge the need for the right to privacy to continue to be upheld in statute, but application of existing statute protection to genetic information is something that was never intended when such laws were passed. Even were it argued that current safeguards could be understood to include genetic information, like all such claims it is subject to legal challenge. Considering the nature of genetic information and the potential damage misuse could cause, legislation specific to use of genetic information proactively introduced before problems arise would be preferable to reactive changes to legislation after the damage has been done.

Courts of law have traditionally avoided making decisions that conflict with established scientific or medical practice, instead relying on medical and scientific practitioners to provide some degree of self-regulation[5]. There are many medical practices established in a past environment where peer review was less rigorously applied, that are questionable in the current ethical environment. Where such practices have been challenged legally, courts are reluctant to take action because of a general lack of understanding of medicine and science. The unfortunate side effect of this is that the judicial, general legal and regulatory environments have fallen behind the scientific and medical environment, particularly in the area of legislative protection and regulation of use of genetic information. To a large degree, enforcement of societal views of ethical behaviour in relation to scientific and medical issues, has moved from the realms of the judicial system to that of peer regulation. Such an arrangement is particularly dangerous where practices are well established as those who do challenge what they consider to be inappropriate behaviour are, more often than not, seen as outcasts because of their views. This not only defeats application of peer review, but prevents or delays changes to practices that are often seen in the wider view of social ethics as being clearly inappropriate. Whilst requiring a carefully considered approach to change, it is clear that consideration of certain practices in the context of current societal standards should be the responsibility of the judicial system, not medical and scientific practitioners. Ideally, legislative regulation of genetic information should be provided by the Federal Government, current arrangements with State legislation providing a piecemeal approach to privacy potentially allow for inconsistencies in application of protection. Codified law has a place and works effectively for legislation of a more general nature, but application in the context of the rate of change currently occurring within the scientific and medical communities would be clumsy at best. In our view legislative regulation at the Federal level, whilst an approach that errs heavily on the side of caution, is clearly needed for protection and use of genetic information, particularly as it relates to privacy.

There is an important distinction to be made here between regulation of institutions and regulation of information. Regulation of the information type, regardless of who seeks access to or use of that information, means that all are bound by regulation. This is especially important give the numbers of commercial concerns seeking access to genetic information. Past situations, such as the application of freedom of information legislation only to medical records held by the public sector, need to be avoided. Whilst this problem has been addressed recently, it has taken a very long time for the message to filter through from those who for so long sought access to their medical records, to those that finally introduced the legislation to make it possible. It should not be the case that privacy legislation for the protection of genetic information has to undergo such a tortuous introduction. For this reason we believe that Federal privacy legislation should be introduced as a matter of priority that regulates access and use of genetic information, not behaviours specific to any institution or group of institutions.

EQUAL OPPORTUNITY AND ANTI-DISCRIMINATION LEGISLATION, DAMAGE CONTROL OR PRO-ACTIVE PROTECTION?

Discrimination hurts everybody. Apart from the personal and social cost, recent damages payments awarded for discrimination cases, have sent a clear signal that discriminatory practices are neither socially acceptable nor responsible and that there is a financial penalty to be paid by those who discriminate. Equal opportunity and anti-discrimination legislation, both Federal and State, describes various attributes on which basis it is unlawful to discriminate.

As societal awareness of various issues increases, so additional attributes are added to those already existing in legislation. This approach has evolved in direct response to increased societal awareness and concern about discrimination on various bases, as expressed in a judicial environment. Attributes added in this way can truly be considered a reflection of societal concern. As an example, those who identify and live as intersex rather than male or female, are now afforded specific protection under the Victorian Equal Opportunity Act – "If you: are intersexual (born with anatomy or physiology different to current ideas of what constitutes "normal" male or female)"[6]

Genetic information is relatively new when viewed in direct comparison to other information that may be used to discriminate against a person. The attribute with which most people associate discrimination, that of being male or female, has existed at least as long as specific vocations have been recognised, possibly longer. When compared in such a way, discriminatory application of genetic information is in its infancy. What is clear, despite only relatively recently beginning to be understood, is the potential for genetic information to be used in a discriminatory manner.

As access to improved testing and analysis techniques becomes a reality, both practically and financially, more information about familial and personal medical histories is potentially available. Whereas other attributes in anti-discrimination legislation are matters of fact that relate to one person and are able to be established at a given date and time, genetic information is able to be applied to individuals and familial groups and gives the impression of establishing future fact. Both of these features unique to genetic information give it a potential for discriminatory application not possible with other forms of information.

Discrimination on the basis of having an attribute not common to wider societal groups has been around for as long as mankind. Social outcasts have traditionally been so because of some physical attribute that made them stand out from their peers, either because that physical attribute made them weaker or because they appeared different to those around them. Some societies have attached special roles to those with differing physical attributes, as an example the special religious or cultural status given to some with intersex conditions in some pacific island groups. In other cultural or religious groups, women are prevented from taking on particular roles. Whether outcast or given a particular role in a social group, the effect of such interaction is to give a degree of separation from their peers that prevents social integration and is therefore discriminatory. The same principle applied to a wider cross section of society, gives us the basis for anti-discrimination legislation today.

Genetic information as a basis for categorisation of people into particular groups is unparalleled. Social division by application of genetic categories would override or subsume virtually all other attributes previously used in this way. The genetic underclass created by divisive use of genetic information could potentially cut through all racial, cultural, religious, gender and economic groups in society.

As discussed previously, the way in which information about a genetic condition is disclosed is an important part of coping with the realities of living with the condition. Not only does discrimination have the potential to cause distress for a person living with a genetic condition, it has the potential to seriously undermine the mental health of that person with the resulting cost to society of medical treatment. Treatment of those with genetic conditions as having “defects” gives rise to feelings of persecution and isolation, discrimination on the basis of having a defect reduces the sense of self worth of a person with subsequent effects on mental health. It is virtually impossible to draw a line between a genetic variation and a genetic defect for any practical purpose, personal experience creating a highly subjective application of this test. To allow discrimination on the basis of genetic variation, and by association genetic information is unacceptable.

Delays introducing something as simple in concept as sex and racial based discrimination acts, resulted in many cases of discrimination that have become frequently cited as case law for future matters. Those who are HIV positive are still openly discriminated against despite legislation that prevents discrimination on the basis of medical condition. Clearly, current “blanket” anti-discrimination attributes that are open to wide interpretation of application are not sufficient to protect from discrimination against those who fall into specific groups perceived to be possessed of socially unsound behaviour. This same attitude is seen in the approach to collection and recording of genetic information from prisoners. Previous behaviours seen by wider society to be unacceptable, have translated to a reduction of protection from discrimination. Like all matters that involve subjective consideration of human rights, where one person considers behaviour to be socially unacceptable, another may not.

Whilst many would not dispute taking DNA human tissue samples from prisoners and the subsequent reduction in human rights this entails, it does not take a quantum leap of imagination to see definitions of what is considered to be socially unsound applied to a wider cross-section of society. Some time ago, there was much controversy surrounding the Victorian medical community’s decision to place lower priority on medical treatment for those who smoked cigarettes. Once again, this is a product of behaviours considered to be socially unsound. Should it be the case that those with genetic conditions are

seen in the same light as smokers? Should someone with a life expectancy that is considered shorter because of their condition, be denied medical treatment as a result? No-one of reasonable social conscience would argue the case for this approach, however, matters of social conscience are frequently overridden by financial or other risk based concerns. It should not be the case that little understood conditions or information about those conditions be used as a basis for discrimination.

Many institutions have taken care to avoid or remove potential areas of discrimination. The International Olympic Committee, deliberately removed genetic testing of applicants for women's events, because they had unwittingly disclosed cases of AIS having discovered women with XY (typically male) chromosome patterns. Not only was disclosure in this manner unacceptable, there is still considerable personal variation in the way women deal with having AIS (some do not even tell partners), but the issue of discrimination against women who are physically no different to their counterparts with XX chromosome patterns was seen to be a serious one. There are many other situations where discrimination might occur without any justification simply because of the lack of understanding of genetic information and genetic conditions.

Refusal to undertake genetic testing or to provide genetic information should not be grounds for discriminatory practices in the fields of employment or insurance. There seems to be emphasis on questions of potential discrimination once information is disclosed, why must it even be disclosed in the first instance? Many areas of law enshrine the right to inaction, including criminal law. Refusal of consent to provide a sample for criminal DNA profiling requires an investigator to make application before a member of the judiciary to obtain the sample, this for criminal matters. How then can insurers and employers possibly justify forcing a person to submit to genetic testing or to dismiss or refuse employment or services as a result? Certain scenarios cited by employers and insurers in an attempt to justify their application for access to genetic information, require highly subjective interpretation of results. The public safety argument, if taken to extremes, would mean each of us having to undergo genetic testing just to obtain a driver's licence. No insurer or employer group can justify statistically the need for access to such private and personal information in the interests of public safety, nor the sort of discrimination they are suggesting as a result of genetic tests.

Without doubt, the underlying principle of equal opportunity and anti-discrimination legislation is the Australian ideal of a fair go for all. In practice, it is most often the case that equal opportunity legislation is one step behind rapid societal attitude changes. An inevitable by product of this "legislation lag" is that some person often has to suffer damage before the law recognises the need for change. The combination of delayed introduction of legislation and insufficient definition of attributes in that legislation has in the past given rise to continued discrimination, something that should be avoided at all cost with the potential discriminatory uses of genetic information. Few would dispute that urgent steps should be taken to safeguard genetic information. In our submission, the safeguard that needs to be put in place is specific legislation preventing discrimination on the basis of genetic information. Ideally no-one other than medical institutions should ever have access to the information in the first instance, but it is imperative to legislate to prevent misuses of genetic information where access is obtained in inappropriate circumstances.

FOR WHOM DOES THE REAL RISK EXIST? INSURANCE AND GENETIC INFORMATION.

Insurance is becoming a subject of considerable public focus. Premiums are rising with insurers claiming that the rises are due to increased claims and the removal of premium "ceilings" that had been kept unsustainably low for too long. Public indemnity insurance premiums are becoming so expensive that many public tourist and entertainment venues have closed through inability to meet the associated costs. In this environment there are increased pressures upon insurers and those they insure, to reduce risk and the related cost of insurance.

Insurance groups are regarded with a degree of mistrust by many in society. With media focus on insurance issues usually directed at disputed claims or refused claims, this is not surprising. Insurance companies are seen as being financial Goliaths, against which some very public Davids have taken up the fight. Collapses of some very large general and specific insurers have served as a reminder that insurance companies do not have access to financial "bottomless pits".

There exists some genuine reason to mistrust insurers though, recent health insurance matters calling into question justification for some actions they take. Largely in response to insurance industry concerns about low membership in health funds and the potential inability to meet costs, the Federal Government introduced a scheme that put a "financial gun" to the heads of most Australians who were deciding if health insurance was a valid option. Predictably, a measurable rise in health fund membership resulted from the scheme. Insurance groups have now applied to Government for an increase in premiums, claiming the increase in membership has resulted in higher costs. Either their financial forecasting methods need overhauling, or they are not being honest about reasons for their application for a premium increase.

It is against this background, that insurers are seeking access to genetic information to help them calculate risks associated with certain types of insurance. Initially, they are seeking this information from those applying for life insurance policies, but as other forms of insurance such as public liability insurance become increasingly expensive, the same pressure will apply to groups seeking to minimise risk and cost as currently apply to life insurance.

The underlying reason insurers cite for applying for access to genetic information relating to policy applicants, is to undertake risk assessments of those applicants. In reality, neither genetic testing technology or statistical modelling of outcomes of physical manifestation of conditions where predispositions exist, have developed to the stage that either will give results accurate enough for such predictions of risk. In addition to questions about positive tests, are concerns related to conditions that may spontaneously occur regardless of a negative test. AIS is a case in point. AIS is linked to the X Chromosome and the gene which causes the condition has been identified and can be tested for, with certain limitations. A mother with carrier status has a one in four chance for each child born being born with the condition and a one in two chance of her daughter becoming a carrier. The condition itself, manifests physically in different ways along a "sliding scale" for each birth with the condition, sometimes manifesting itself differently amongst siblings in one family. In those where the effect of the condition is less obvious, it is virtually impossible to detect any genetic evidence of the condition. Spontaneous manifestation of the condition also occurs, so in a family pedigree where the condition has

not been historically present, it may appear without prior indication. Even when the condition is genetically indicated in an unborn child, there is no way of predicting the degree of physical manifestation until the child is born.

Basing access to insurance on outcomes of genetic testing also threatens to deny access in some cases to those who need it most. Discrimination against minority groups with genetic conditions so that the majority may enjoy lower premiums, is socially irresponsible and could be considered indicative of a general lowering of social services. To reduce Government based social services and increasingly rely on the private sector to replace those services on one hand, whilst on the other hand allowing providers of those services to apply them in a discriminatory manner or place them out of financial reach of those who need them, has the potential to create a genetic underclass problem never before seen.

Whilst as a general rule, Australia is a world leader with such things as prevention of discriminatory application of the law, the United Kingdom model of applying a benefit ceiling to life insurance policies before genetic testing is requested would seem to be a fair approach to the insurance problem. As long as such ceilings are applied in such a way that makes them subject to indexation against such things as increases in average wages, they have the potential to ensure access to such policies by all who need them. As a general rule, we believe that insurers should not have access to genetic testing. If premiums rise as a result of ensuring all who need services are covered, then that is the price of being socially and morally responsible.

"VOCATIONAL GENETIC TESTING", WHAT ARE THE ALTERNATIVES?

Use of genetic information by employers would fail the test for use of genetic information we have outlined herein as it is not beneficial to anyone that could be described as being a "patient". Employers might argue that they have genuine reason to have access to genetic information where a perceived susceptibility to a condition exists in a field of employment, or vocations where public safety risk concerns exist. This latter issue is becoming more of a concern because public indemnity insurance premiums are becoming impossible to afford in some situations. But does an employer have to have access to genetic information to adequately gauge these risks, and furthermore, does genetic testing provide results accurate enough to use in these circumstances?.

As outlined earlier in this document, it would seem that it is often the case that misconceptions about the possible use of genetic information abound. Short of specifically employing medical professionals solely for the purpose of conducting what we would describe as "vocational genetic testing", it is unlikely employer groups would be able to adequately interpret the results of genetic tests in any reasonable manner. By reasonable manner we mean without adopting an unnecessarily exclusive use of the information elicited when gauging suitability of a particular employee or applicant for a position. It is currently the case that medical practitioners or scientists are largely unable to convert, with any degree of accuracy, identified pre-disposition to a condition into actual likelihood of manifestation of the condition in any physical sense. It follows that employers, even basing decisions on medically interpreted vocational genetic tests, are likely to exclude from employment on the basis of genetic pre-disposition to

a condition a percentage of prospective employees that will never ultimately be affected by the condition to which the pre-disposition exists. Would an applicant, excluded from employment on the basis they are genetically pre-disposed to a condition, have a case for legal action against that same employer upon retirement if the condition ultimately did not manifest itself?

There is, notwithstanding the above concerns, no arguing the case that any person excluded from a field of employment in which they are particularly susceptible to harm, could be so excluded on genuinely humanitarian grounds. Is there a way of striking a balance between the needs of the prospective employee and an employer wishing to minimise workplace risk? There would likely be no objection to having an applicant undergo vocational genetic testing at the request of their GP. The results of such genetic testing could be explained by the applicant's GP (or chosen genetic specialist), in context of the risks in the field of employment sought. Employers could prepare documents, to be signed by an applicant and their GP as having been discussed, that outlines the risk to employees with certain genetic pre-dispositions. Whilst such "general indemnities" as these have been challenged in other insurance and recreational related applications, there is a sound argument for a document that places the duty of care for health related matters back where it belongs, medical practitioners who have a duty of care to their patient and who should be acting in their patient's best interest. Such a procedure keeps sensitive information away from employers, information gained as a result of any vocational genetic testing being known only to applicants and appropriate medical practitioners. There are certain types of information that are clearly irrelevant to employment prospects, these would be safeguarded by such an approach.

Seeking medical information from current employees of long standing in an organisation is also another issue that needs regulation. Approaches such as these to current employees could easily be seen as a way, especially for older employees, of looking for a way to terminate employment and retain a younger workforce. Statistically speaking, the older an employee is the greater chance there is of them having a medical history containing information that might be used as an excuse to terminate employment.

A Federal Government agency recently sent a message to all employees seeking full disclosure of medical histories and requesting that some employees have examinations by their doctors. In reality, the concerns they are seeking to address could easily be dealt with by asking for a certificate from the doctor of each employee stating they are fit to be employed in the area they are working. This would save employees having to provide information to their employer they would rather keep private and provides sufficient protection for the employer. This is a good example of the way alternate approaches to employment issues surrounding disclosure of genetic information might work.

Protection of genetic information in the manner we have described, safeguards not only applicants records but also employers from any accusations of unlawful disclosure to other parties. Whilst we believe that any perceived advantage to vocational genetic testing is outweighed by the potential discrimination that may result from such an approach, we would rather see testing conducted in the above manner than by employer groups themselves.

MEDICAL PRACTITIONERS, GATEWAY TO THE FUTURE OR BARRIER TO PROGRESS?

General practitioners are the gateway to medical services. In all likelihood a person seeking medical assistance will approach their GP for assistance in the first instance. Direct approaches to other specialists will generally result in a request for referral from a GP. It is important therefore, that GPs have a good understanding of what genetic information is, what it may or may not indicate, how to go about obtaining genetic information and how to explain results of genetic tests to a patient in their care.

Like any group that deals with genetic information, GPs must be appropriately educated and have adequate training if they are to use the information effectively and responsibly. It is an important part of the process of disclosure of the existence of a genetic condition, that the person disclosing the information is trusted and respected by the person being given the diagnosis. Understanding the information and what it really means for their patient is also vitally important, as there is much misleading and inaccurate information about genetic conditions. GPs are also going to be very reluctant to refer patients for genetic testing or to genetic specialists if they are not comfortable using the information that is an end result of the process. " General practitioners reported that management of genetic conditions was an infrequent part of daily practice and they learned about rare conditions when they needed to. Very few had any experience in ordering a genetic (DNA) test and most had limited experience in referring patients to genetic clinics or to support groups".[7]

GPs should also be comfortable safeguarding genetic information. As a general rule, GPs have a good reputation amongst consumer groups for protection of patient information. Such a reputation could easily be destroyed without appropriate guidelines for recording and storing genetic information on patient records along with knowledge of appropriate disclosure upon referral to specialists. GPs should not be placed in a position where appropriate handling of genetic information is a matter of guesswork. Medical associations should work with consumer groups to provide guidelines for the handling of genetic information by GPs.

Learning to involve a genetic counsellor in the disclosure process is also very important. As mentioned earlier in this submission, genetic information is a very persuasive and highly likely to influence decisions about treatment options and such things as terminations of pregnancies. A team based approach, involving a patient's GP and genetic specialists co-ordinated by a genetic counsellor should become standard practice given the constantly increasing incidence of disclosure of genetic information to patients.

THE "FORENSIC PROCEDURES ACT", EFFECTIVE LEGISLATION OR SLEIGHT OF HAND?

The debate of the right to privacy versus access to information in the public interest for law enforcement, has been around since the first formally recognised policing agencies. These often competing interests have been examined by courts on numerous occasions, with outcomes as varied as the situations presented for consideration. Like many non-medical institutions, law enforcement agencies have sought access to human tissue samples as improved testing techniques allow greater accuracy of results. Law enforcement agencies principally use samples for suspect identification and elimination purposes where the apparent “uniqueness” of the information is useful, but genetic information is also used by some law enforcement agencies for research. For most law enforcement agencies, Part 1D of the Commonwealth Crimes Act 1914 is the instrument via which they obtain and use human tissue samples.

All information obtained by law enforcement agencies is still legally held to be the property of the person from whom it was taken, unless illegal to possess and therefore subject to seizure and later destruction. It is also the case that information obtained in this way is subject to strict privacy controls regarding the release of information to a third person and the way the information may be used. The exception to this rule seems to be genetic information obtained by law enforcement agencies. Whilst most people would support use of genetic information for law enforcement purposes it is clear that regulation of such use is as important as it is for other institutions currently using information obtained from human tissue samples.

A clear pattern of law enforcement collection and use of human tissue samples obtained via the Forensic Procedures Act is emerging that should be carefully monitored and regulated. Human tissue samples obtained for law enforcement purposes are originally obtained with a single purpose in mind, indeed a legislative requirement of their collection is that the purpose for which the sample is obtained be explained to the person from whom the sample is sought. Upon refusal of consent, application must be made to a judicial officer to obtain a sample and, once again, a clear statement of the purpose for which the sample is sought must be made. It should be the case, as with other forms of information used by law enforcement agencies, that genetic information can only be used for the purpose for which it was legally and expressly obtained.

A law enforcement specific issue, is the destruction of samples where legal process is not instigated against the person from whom a human tissue sample is obtained, or where the sample is provided for “elimination” purposes. It is currently the case that samples, once the specific purpose for which they were provided no longer exists, are to be destroyed. This is made clear upon application for consent to take a sample. It is in the definition of destruction that the issue exists. Tissue samples due to be destroyed are merely “de-identified” not destroyed, and would then be available for use for other purposes, such as research. As discussed earlier in this submission, the so-called “de-identification” of samples is only possible in an abstract way as the sample is still unique to the person from whom it is obtained. Of concern, is the fact that the term destruction is likely to be taken by most to have its ordinary meaning, including the person giving consent for the sample to be taken in the first instance. In our view this definition of destruction amounts to a deception by omission and samples subject to destruction should be physically destroyed. Physical destruction is the only way alleviate the temptation to later use human tissue samples for purposes not originally consented to.

Furthermore, there are questions about the legitimacy of law enforcement agencies conducting research using human tissue samples. Leaving aside for the time being questions about the focus of such research, consideration of such research by HREC must take place as with any other institution. It is very likely that any research that, as an example, might look for common genetic precursors in those that commit certain criminal offences, would fail by any reasonable test to be approved by an HREC. If law enforcement agencies are to conduct research using genetic information, they must be called upon to apply for and account for such research in the same way any other institution must.

It has been the case in the past, that assumptions have been made about the frequency of commission of offences amongst those born with differing chromosomal make-ups, unfairly adding to the stigma already existing for many that have a genetic condition. One group even went as far as to change the name their condition is commonly known by as a result of specific outcome based research. Not only would research that looks at these sorts of questions very likely fail to gain approval from the relevant HREC, it is ethically questionable at best, at worst a return to the sorts of unchecked research conducted in very inappropriate circumstances in the 1920's and 1930's.

Informed consent is an issue considered in detail earlier in this submission. The same questions about informed consent apply to collection and use of human tissue samples for law enforcement purposes as do for research or other purposes. In short, asking a lay person to consent to the use of information about which they have limited understanding is ethically questionable, especially where application for collection of samples may be heard ex-parte. Given that there is an alternate process in place, that of going before a member of the judiciary to make application to obtain samples, it is not unreasonable given the type of information concerned to have all requests for samples dealt with in this manner.

A final issue regarding law enforcement use of human genetic information is access to information obtained by the medical profession for a medical purpose, either by hospitals as a result of general screening or by specific testing. There is always the question of public interest versus privacy when considering giving law enforcement agencies access to information obtained originally for a non-law enforcement purpose. Regulation of this has become so strict that most organisations now require service of search warrants before they will disclose information to law enforcement agencies. Whilst many law enforcement agencies may consider this to be frustrating in some instances, the need to protect privacy has become paramount given the multitude of ways it may now be invaded. We believe genetic information obtained for medical purposes, because of the amount of information about a person it reveals, should be "legally quarantined" from law enforcement access.

Evidentiary issues related to the introduction of evidence regarding DNA "profiling" or "matching" are something that the forensic science community and courts will have to address. Short of introducing special tribunals to hear and decide matters related to scientific evidence and an abolition of the principle of a person being tried by 12 of their peers for at least that part of the trial process, the only other way of dealing with inadequate understanding of the limitations of genetic information used this way is greater community education. It is very unlikely that a jury, even one considered to be educated and instructed appropriately, will understand in such a short time the nature of genetic information and the weight it should be given in a tribunal of fact.

A CLOSING REMARK.

A Queensland MP once commented that genetic information obtained as a result of anti-natal screening of infants should be placed on the NCIC database because "no-one has anything to hide if they do not commit a criminal offence". Simplistic views such as these demonstrate the complete lack of understanding of the nature of genetic information and the way in which it may be used and is indicative of the general level of misunderstanding about such issues. It is imperative that those who seek to use genetic information for any purpose and those who would legislate to protect or regulate the use of genetic information, do all they reasonably can to ensure widest possible consultation so that their level of understanding of the issues involved is sufficient to protect those to whom the information belongs. Never before has a science that offers so much to all people, had the potential to be misused to so great a degree.

NOTES:

[1] Tissue that contains both testicular and ovarian tissue.

[2] "Beyond Two Sexes" Ian Jackson/Billy Waters. Pub. New Scientist No 2290 12 May 2001 (pp 26)

[3] "Beyond he, she and it". Anthony R M Briffa, President AIS Support Group Australia. Pub. New Scientist No 2293 2 June 2001 (pp 54)

[4] Report on a study at the Imperial College Of Science, Technology and Medicine of outcomes of disclosure of genetic information to parents. By Dr Lenore Abramsky and Ors. Pub. British Medical Journal, 14 February 2001; (322; 463-466)

[5] "An Emerging Ethical and Medical Dilemma: Should Physicians Perform Sex Assignment Surgery on Infants With Ambiguous Genitalia". Hazel Glenn Beh Ph.D. and Professor Milton Diamond Ph.D.

[6] Victorian Equal Opportunity Act 1995 as amended 9 October 2000

[7] "Getting the gene into general practice". Jennifer Newstead, MBBS, FRACGP and Sylvia Metcalf, PHD; Pub. Australian Family Physician Vol 30, no 10 October 2001 (pp 927), from a needs assessment of rural and metropolitan GPs by RACGP Victoria, the Murdoch Children's Research Institute and the University of Melbourne.