Portuguese General Report

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I – Legal status and general rules concerning genome analysis and its application in general

There are, in Portugal, no specific rules concerning genome analysis. This matter is too new to have been broadly discussed in our country.

Genome analysis, which is practised, as we shall see, in some domains, is governed by the common legal system that rules all medical activity and forensic medicine.

At the highest level of our legal system we can find some basic rules in the Constitution of the Portuguese Republic.

Article 25 deffends individuals against any offense to their physical or moral integrity. This rule, well known in Portugal as in many other legal systems, is the legal basis for the necessity of respecting individuals' autonomy in two distinct ways: firstly, it demands the obtaining of an informed consent prior to any practice of medical intervention regarding genome analysis; secondly, it reserves for the individual all power to decide about the knowledge and disclosure of results of genetic testing.

Article 26 of our basic law expressly protects privacy. In spite of its vagueness and imprecise limits, nobody doubts that it keeps individuals secure against any non authorised intervention on their body, conducted by private physicians or by state services, regardless of the purpose of the intervention.

Still at the Constitutional level, article 32, n.6, establishes that all evidence obtained in a criminal process, in a way that offends physical or moral integrity, or involves a non authorised intrusion in private life, is void. And the same rule is established in the Code of Criminal Procedure (Art. 126, n.2/3).

In this description of general rules of our legal system dealing with the respect of autonomy, I must add the precept of the Penal Code that punishes, as a criminal offense, any medical act performed without the consent of the person concerned (Art. 158).

Another law of great importance – the Civil Code – has a specific rule to protect privacy (Art. 80) and, in a broadly formulated rule, deffends individuals against any kind of offense to the basic aspects of their personality (Art. 70).

Our basic law also protects, with particular strength, the right to personal privacy. Article 26, n. 2, orders that ordinary statutes enact rules against non authorised disclosure of data concerning identified persons and families. And article 35, n.3, forbids automatic processing of personal data. (I must notice that this rule, so strictly formulated, could be understood in a very inconvenient way, creating an obstacle to the organization of automatic data processing in hospitals. But this has never actually happened).

At the level of ordinary statutes, there are several laws on professional duty of confidenciality. For instance. the Regulation of Labour Medical Services 1967 (Art. ''1) and the main law about Labour Contract 1969 (art. 124).

Last October. a law about protection of personal data was published (Law n.10/91). The law is about protection of all personal data and not only medical information; but it is quite clear that the legislator had in mind automatic medical files. Article 32 outlines the duty of secrecy that obliges all those responsibles for storage and use of personal data. and Article 41 reinforces punishment of misuse of information by doubling the penalty that was established by the Penal Code.

Beyond these regulations, we must underline that Portuguese physicians and health administrators have a long tradition of confidentiality and a clear awareness of the necessity of keeping secrecy about personalised medical data. No one responsible for a medical file would disclose information without the permisson of the person concerned.

There are good reasons to expect some increase of medical liability soon; but those conflicts will probably have nothing to do with lack of confidentiality.

II – Labour and insurance law

1. Labour law

As expected the Portuguese rules of labour law and above all those concerning health in labour, don't mention genetic tests expressly.

However, the general rules in force are formulated in such an extensive way that they are competent to frame, without any effort, every progress in the sanitary practice of the company.

A regulation from 1967 did already impose the performance of admission and periodical tests to workers, with the aim of controlling their aptness for holding a position and of verifying " the repercussion of the work and its conditions on the workmen". Obviously at that time no one thought about genetic tests or about presimptomatic diseases; but the legal formulation was already sufficiently flexible and capable of embracing them.

More recently, in a law from November 1991 that intended to adjust the national law of the Portuguese state to Directive 89/391/EEC, it was established that the companies are obliged to organize, at all levels, the evaluation of the risks concerning the safety and the health of the workers" (Art. 8, 2, b); the employer is obliged to "assure that the exposure to chemical, physical and biological agents at work, does not set up any risks to the workers' health"(c);furthermore he is also obliged to "assure the adequate vigilance of the employees' health considering the risks that they are exposed to at work" (h). Lastly, we expect at any moment the promulgation of a regulating diploma whicht should, following Art. 23. 2, d) of the 1991 law, contain rules about "groups of workers who are specially sensitive to certain risks, namely youth and pregnant women".

It might be said that in these laws, as in some others connected with the admittance to the Public Services, or with the carrying out of certain occupation that demand special sanitary care (e.g. the nourishment industry), the Portuguese legislator always had in mind the physical aptness of the worker, the checking of absence of contageous diseases, and was always specially careful with pregnant women and youth. But it can also be said that the more recent legal texts refer expressly *all kinds of risks* that could be motivated by the permanence at the workplace; and *hypersensitive groups* are taken into account besides pregnant women and youth.

This judicial frame, that was established in general for the whole Labour health system, allows all companies to consider the performance of genetic tests to applicants or to workmen on duty.

There are however several reasons that demand great caution in relation to the performance of genetic tests and with the consequences that might follow them.

First of all, the circumstance that several genetic diseases are of eventual or late development, in such a way that it can't be taken for granted that the worker is at a particular moment ill.

Secondlly, the fact that in many cases no rigorous connection can be established between the manifestation of the disease and a specific circumstance of the workplace should also be taken into account. And lastly, it is disturbing to think that DNA techniques will provide much more information than that which is aimed at in the actual case, with the added risks of a breach of confidentiality.

Under these special conditions, it seems desirable to demand the verification of supplementary requirements for the performance of genetic tests:

a) That the relation between the worker's hypersensitiveness and the actual conditions of the workplace be clearly defined;

b) That the inquiry be made exclusively in the interest of the sanitary protection of the individual submitted to the tests or in deffence of the integrity of a third party, which cannot be deffended otherwise.

These considerations, however, do not deal with the problem of determining whether the tests may be imposed on or merely suggested to employees by employers, leaving to the former the autonomy of decision. Two basic solutions are admitted: either the tests are imposed or they are only suggested – in this case, the employee is free to decide whether or not to submit himself to the tests, but if he refuses, the employers are exonerated from the financial responsibilities that would otherwise be theirs.

In the former 1967 Regulation it was clearly stated that employees "could not exempt themselves from" sanitary tests (Art. 15); however, the 1991 Law in force establishes only a general duty to "comply with the prescriptions concerning (...)Labour health established in legal dispositions" (Art. 15, n. 1 a)).

I think that a system based on simple counselling may be suitable in culturally advanced countries, with a good *per capita* income, great social mobility and an effective social security system, where the employee can seek counsel and where the choice of a different type of activity involving no health risks is actually available. The "sanitary paternalist" imposing solution might, on the other hand, be suitable in those countries where it can be expected that candidates, in a situation of necessity, will despise all advice and refuse any test that can put at risk the possibility of present work, whatever eventual health damage they may experience in the future. In this case it might be adequate to have compulsory tests.

However, the consequences for the candidate of a positive result in the tests he was submitted to under the latter system should be carefully balanced.

I do not think that we should adopt the easiest solution of immediate exclusion of the hypersensitive candidate or dismissal of the worker whose presence in the workplace puts his health at risk. Before legitimizing the decision proof shoud be required that it is not possible to eliminate health risks by changing the place of work or by introducing technical improvements; or that, though technically possible, those improvements would be economically disproportionate. I believe that a balance of this type might comply with the deffence of the workers' individual rights, with the need to render the workplace healthy for actual and potential workers and with the necessity of not imposing too heavy an onus on employers.

I would like to stress another aspect that should be carefully regulated: the preservation of test results.

According to the norms in force in Portugal, the confidentiality of data is garanteed and the right of access is granted only to official inspection services. The potentially more informative character of tests based on DNA techniques suggests the need for a reinforcement of caution to garantee confidentiality. It is doubtful that it will be possible to maintain a norm similar to the one found in the 1967 Regulation (Art. 21, n. .2) stating that the worker had, when leaving the place of work, the right to claim "a duplicate of his medical file", but not the original.

These considerations reflect my own opinion: this type of sanitary control of the workplace has not yet been discussed in Portugal.

2. Public insurance (social security)

Social security rules do not refer genome analysis as was to be expected.

But national screening of Phenylketonuria (PKU) and Hypothyroidism is paid for by public health systems as well as the therapeutics used in detected cases – e.g. diet in cases of Phenylkenoturia.

The circumstance that new borns were voluntarily submitted to "screening of metabolic diseases" is mentioned in the child's Health Card, for which the Ministry of Health is responsible. It might become clear whether payment or a subvention are always garanteed by the State when children were not submitted to screening at the right time.

Expenses with Familial Amyloidotic Polyneuropathy (FAP) are also entirely covered by public social security. We could say that those patients enjoy a privileged position in this respect: expense coverage includes not only a large amount of medical substances and equipment but also the personal following up that patients need at the later stages of the disease. This assistance is not dependent on the fact of the diagnosis having been timely or of families having followed medical advice in the organization of their lives, namely where marriage and procreation are concerned.

The same applies to inquiries that do not reach the dimension of a screening and to the therapeutics or the folowing up of other less frequent diseases: Haemoglobinopathia, Haemophilia and Cystic Fibrosis; also to chromosomal testing for detection of the Down Syndrome.

3. Private Life Insurance

In Portugal, as in other European systems, life insurance contracts are based upon a declaration by the insured about their sanitary history. This declaration is taken as true by the insurer, who does not impose the performance

of any medical exam in cases where the amount implied in the insurance contract is considered normal.

If the amount involved is higher, insurance companies require progressively more exams: e.g., if the amount is over 25 million escudos the list of clinical tests imposed will be considerably long.

However these tests do not use DNA techniques and are not therefore addressed to the detection of genetic diseases. This does not prevent them from detecting diseases with a genetic component, such as diabetes. This case has been incidentally taken into account for a long time by insurers who translate diabetes into a shortening of life expectancy, thus demanding from the insured a higher premium.

Given the present state of development of medical genetics, Portuguese laboratorial conditions and specially the economic situation of insurance companies in Portugal, relevant or rapid modifications are not to be expected in this matter. Private insurance used to be very defficient until a few years ago; nowadays there is a tremendous expansion in the activity of insurers accompanied by enormous competition. These conditions seem to be leading insurers to concentrate mainly on increasing the number and volume of contracts and not on too rigorous a screening of candidates, which might prove expensive and socially unpleasant.

In a future I cannot for the moment foresee, it is to be expected that insurers will include in their demands the detection of genetic diseases, at least of those whose testing be relatively inexpensive and well known in Portuguese laboratories.

I do not know of any discussion of these matters in Portugal. But the choice of the legislative attitude will probably be made from the following posibilities:

a) Insurers shouldn't have the right to require genetic testing or to inquire about results of previously performed tests, as a precondition for the conclusion or the amendment of an contract;

b) Insurers shouldn't have the right to require genetic testing, as a precondition for the conclusion or the amendment of an insurance contract; but they should have the right to ask the applicants for the result of previously conducted tests that have been made for any reason other than insurance;

c) Insurers should have the right to demand of the applicants, as a precondition for the conclusion or the amendment of an insurance contract, either the disclosure of previously conducted tests made for any reason other than insurance, or the submission to genetic tests that, on the grounds of serious and present indications, appear as expedient to make an informed judgement about the health of the applicants;

d) Insurers should have the right to demand of the applicants, as a precondition for the conclusion or the amendment of an insurance contract, either the disclosure of previously conducted tests or the submission to genetic tests, whenever it will prove adequate to make an informed judgement about the health of the applicants.

It might be reasonable to argue that it will be difficult to deny insurers the resort to *special means* in case of contracts of *special value*. It would therefore probably be fair to admit that insurers be authorized to know the results of tests previously conducted in competent medical institutions; it seems less acceptable to allow new tests to be conducted as a condition for the conclusion of a contract. unless there be very serious evidence that the candidate suffers from a genetic disease that he knows or should reasonably know of.

If the contract is within the *normal values* practised in that branch of activity it does not seem acceptable that insurers may demand submission to any test or the disclosure of previously conducted tests as a condition for the conclusion of the contract.

The economic activity of insurers is based upon an estimate of risk distribution and the probabilities of verification of the damages. If it were possible to foresee the damages exactly and to eliminate the candidates with a strong probability of becoming onerous to the company, insurers would actually eliminate risk – the very basis of their own activity. Neither can it be said that the victims of genetic diseases will ruin insurance companies since these companies already have the charge of paying insurances of people affected by genetic disturbances and do not therefore acquire a new charge simply by not being allowed to exclude these bad clients.

On the other hand, the simple rejection of candidates who carry genetic anomalies creates what has been called "the new class of the uninsurable", who are further sacrificed as the State withdraws from activity in the area of social security.

Besides, the systematic demand for genetic tests could cause the neglect of the imperative need for medical counselling. And it would be an extreme violence, because of a common insurance contract, to make a serious or even mortal disease known ten or twenty years before the development of any symptoms!

These restrictions should not however eliminate the basic rule on which the insurance contract stands – the declaration of good faith made by the candidate. In these terms, the moment the insurer is called upon to pay the insured capital, even when the contract involves a common amount, he should have the right to question the insured and those who detain his medical file on whether,

at the time of conclusion of the contract, the insured knew or should have known that he suffered from a specific disease that he omitted from his declaration and that was later revealed. It seems convenient that this inquiry be made by decision of a court and only when the court thinks the inquiry is justified. The protection of the privacy of individuals cannot justify bad faith; and insurers, in spite of their social function, should not be forced to tolerate any lie. Besides, the Commercial Code establishes that all omission of relevant and known facts by the insured determines that the insurance contract be void (Art. 429).

4. Private Accident Insurance

Private accident insurance does not present any specificity in relation to preceding subjects.

We could however say that this is the field where insurers are used to obtain clinical information about the insured. As a matter of fact, following accidents, insurers frequently request that the court demand medical information concerning the limits of the insured's incapacity resulting from the accident in order to fix the amount of the amends.

I think this is a reasonable way of proceeding and that it will not be extended to the fields that we are dealing with.

5. Private Illness Insurance

The preliminary considerations made in relation to private life insurance apply basically to private illness insurance. That is to say, insurers are in a period of expansion of clients and do not disturb this growth with particular requirements such as genetic tests.

Besides, private illness insurances are expanding as group insurance, and this precludes specific inquiries into the health of each member of the group.

On the other hand, the lower prices and the greater number of insured will allow insurers a better distribution of the costs caused by the insured that will suffer from genetic diseases.

Finally, insurers will be able, in many cases, to share with the State the charges of patient assistance.

For all those reasons this will not be the field where insurance companies will be the most aggressive in their attempt to know more personal data. And it is a field where it seems more acceptable to onerate the companies more than the strict rules of Law might recommend, considering the social function of insurance companies and the dangers for the individuals concerned of the disclosure of these types of disease.

I do not however think that the principle of good faith in the conclusion of contracts should be abandoned; I would therefore admit, in favour of insurers, that they require from the court information about the insured's attitude at the time of conclusion of the contract.

III – Medical law and procedure

1. Medical application

All medical activity related to genome analysis is performed in special, officialy authorised and well prepared institutions. And all medical acts performed are under the responsibility of qualified physicians, though some laboratorial procedures are develloped by other specialists (biologists, chemists).

All genetic testing or genctic screening, in the medical field, is conducted after previous informed consent of the person involved.

Acts of genome analysis are free of charges.

a) Genetic councelling

aa) Prenatal

Several medical centers in our country develop prenatal diagnosis of hereditary diseases. When diagnosis reveals a positive result, a risk for the new born to suffer from a genetic desease, Portuguese law permits abortion.

Article 140, n.1, c) of the Penal Pode authorises abortion if, during pregnancy, it can be proved that the new born will suffer from a severe disease or defect of an incurable nature; abortion has to be performed in the time limit of sixteen weeks after conception.

The first technical problem that makes things difficult for doctors is that the period of sixteen weeks is too short, because technical performance of tests demands more than sixteen weeks in almost every case.

Another difficult matter derives from the fact that many genetic diseases are presimptomatic; the carrier of a genctic disturbance can perfectly live for many years before any symptom of the disease appears; it can even occur that simptoms never appear. In these particular circumstances, it can be difficult for a geneticist to assure that the foetus will probably suffer from an incurable disease that justifies abortion.

The typical Portuguese case of the Familial Amyloidotic Polyneurophaty (FAP) is a clear example of this difficulty. It is an incurable disease, without symptoms before the carrier is about thirty years old; almost every carrier of the genetic mutation will develop the disease; and it is a fatal disease in one hundred per cent of the cases. Can it be reasonable to affirm that a foetus carrying the genetic deffect fulfills the requisites of article 140 of the Penal Code? I have been told that in one European country (the second major European focus of the disease) where the symptoms appear about fifteen years later, abortion is denied.

This particular case becomes even more troubling when we take into account that there is a strong hope of getting a cure, sooner or later.

bb) Postnatal

There is a national screening program for diagnosis of genetic diseases in new borns – Fenilcetonuria and Hypotiroidism.

The laboratorial work is performed only in one Institute, in Oporto, and the follow up of positive cases is distributed by several medical centres, for a better geographic coverage.

It is interesting to notice that the screening started ten years ago and was preceeded and supported by strong publicity in the media; in 1991, this national program screened 96.6% of all births.

As you all know, the therapeutics are easy and pose no ethical dilemmas.

The diagnosis of Familial Amyloidotic Polyneuropathy (FAP) and related counselling have been developing in our country.

Counselling related to this incurable and letal disease poses great difficulties to doctors. At the begining, diagnosis and counselling were performed on demand parents who presented their children for observation. But positiver results in tests created great anxiety in families without any hope of cure, and nowadays diagnosis and counselling are made only on demand of individuals aged eighteen (the legal majority). It is useless to do anything before this age; it can be useful to make carriers aware of their situation at the age they start to think of having their own children.

Follow up studies of this particular and dramatic reality in Portugal are able to show some interesting data.

As a first conclusion, responsibles estimates that 90% of persons at high risk demand genetic testing; only 10% refuse to know if they carry the defective gene. The second conclusion is quite peculiar: carriers keep marrying and having children they know will die in a great suffering, ignoring all efforts of genetic counselling.

A brief final note: there will be hard dilemmas in the near future because it will be necessary to select patients to benefit from the two rare terapeuthics that are able to stop the progression of the disease: liver transplant and a special kind of blood filtering.

The problem of genetic counselling—or the absence of it – raise the issue of the so called; Home tests".

In Portugal everyone can buy, in a Pharmacy, a simple kit to search for Glycemia, ignoring any medical councelling.

I am aware that some wise people are affraid of abuses coming from powerful chemical and pharmaceutical industrie. In fact, the easy revelation of severe diseases could be desastrous for the individual.

Personally I think that there shouldn't exist a legal regime of total prohibition or complete allowance concerning this subject, as I have already proposed in the ambit of the workshop in the "Ad Hoc Commitee of Experts on Bioethics" (C.A.H.B.I.), at the Council of Europe.

As a matter of fact I believe that the regime should balance each kind of "home test", permitting the free commercialization of those that aren't susceptible of causing a special anxiety to the individual, that only show mild and ordinary diseases and that might suggest to the individual advantageous forms of behaviours, such as a more balanced nourishment. I'm thinking of tests that accuse presence of sugar in the blood, or any other kind of disease of a low degree of dramatization, that won't cause more embarassment than the revelation of an unexpected pregnancy.

2. Procedure

a) Criminal procedure

There are no special rules concerning the use of tests of genetic nature in the penal process. In other words, these inspections are allowed in the same terms others are.

The examination of people and scientific inspections can be ordained by the judicial authority either officiously or on demand (Art. 154 Code of Penal Process), considering as the "judicial authority" the judge, the prosecutor, depending on the act and according to their legal capacity (jurisdiction) (Art. 1, n. 1, b).

The judicial authority shouldn't order the performance of tests and inspections, which could offend the physical or moral integrity of people (Art. 126). Without harming the respect for this fundamental right, our penalists agree

peacefully that tests on blood or on other biological products can be ordained for the purpose of scientific research. Some people think nevertheless that the law should foresee expressly the licitude of blood tests to avoid any eventual and possible objection that these kind of tests could offend the physical and moral integrity; or offend more specifically a "right to self-determination on information", which reserves to each person his own competence to decide about the time, the range and the purpose of the divulgation of his personal data. But it can be declared that the widely dominating opinion is that a specific rule to legitimate the judicial authority to impose blood tests is not needed.

Indeed in our law there prevails the generally accepted limit that such tests shouldn't be ordained, unless they are considered to be useful for the disclosure of the truth.

It has been also peacefully agreed that once a test has been ordained, no one can be excused from being submitted to it; one can even be obliged through physical force. It has to be added that this obligation of being submitted to the ordained probes aims obviously at an accused or at least a suspect; this obligation cannot be imposed on someone under the pretext of getting a better instruction of the cause.

In the matter of acquisition of evidence, according to the general system in force, the accusation and the defence dispose of the same means (Art. 154 and following C.C.P.). Both can apply for a new inspection. The best possible preservation of the object of the inspection is also foreseen, in case that it has to be repeated.

There are several services, in this legal frame, that provide the task of making blood testing – the Forensic Medical Institutes and the Laboratory of Scientific Police. These official laboratories perform regularly, it might be said, tecniques of DNA analysis. with the usual efficiency of these kind of tests.

I don't know any refusal to voluntary submission to these tests.

It might be considered as a problem of process, at this time, the question of knowing if a genetic diagnosis that shows a severe illness can sustain, for eugenic reasons, the exclusion of ilicitude in a criminal volontary interruption of pregnancy (Art. 140. nl, c - Portuguese Penal Code). The problem arises from the fact, that a genetic desease might reveal itself many years after the birth, in such a way that it may not be easy to declare during the pregnancy that it is previsible "that the unborn is going to suffer, in an incurable form, a severe desease or deformation. . .". The foreseen exlusion of iliciude has certainly considered the cases, where a severe desease, or an existing or developing deformation is detected through fetal diagnosis; but not how a genetic desease of a future or eventual eclosion can be compared to the clearest cases that the legislator had in mind.

b) Paternity suits

The Portuguese processual law has always allowed, in general, the use of inspection tests. In paternity suits that general admittance took the traditional form of ordinary blood tests.

In 1977 the reform of the Portuguese Civil Code intended to follow the international movement in order to make an agreement between the individual's juditial condition and the biological reality of his birth. Thus, Art. 1801 of the Civil code in force is more explicit than ever in admitting every kind of scientific inspection – I quote: " Blood tests and all kinds of scientifically confirmed methods are allowed in order to obtain evidence concerning paternity suits".

In this legal frame there are no doubts that genetic tests can be used in paternity suits.

In practice, however, Portuguese laboratories which perform research in forensic issues do not make a systematic use of DNA analysis for this purpose. The traditional haematological tests attain reliable results, specially since they were reinforced by histocompatibility tests; thus there was no further occasion for making a systematic use of DNA analysis in this matter.

If genetic tests are performed in the ambit of paternity suits, they will certainly follow the general rules of the civil process, and according to these, everyone has the duty to co-operate in the disclosure of truth (Art. 519 Civil Process Code). The unjustified refusal is therefore illegal and the law determines the application of a fine; the judge of the cause has the power to take the probatory consequences he considers to be appropriate regarding the refusal. However, contrary to what happens in the ambit of the penal process, nobody can be submitted to the tests through physical force.

The circumstance of discussing interests of family nature in these proceedings concerning public interest, contrary to the typical proceedings of patrimonial civil process, where mere private interests are discussed, could tempt the doctrine to deviate these proceedings from the regime which is valid for the ordinary civil process, and approach them to penal proceedings, with the aim of permitting a physical compulsion of the pans of the process who through an arbitrary refusal frustrate the processual acquisition of conclusive elements for the veredict and for the constitution of the plaintiff's civil status. However, the individualistic Latin inheritance does not allow this deviance from the general rules of the civil process.

In spite of the fact that statistics of the courts show a figure of 45% of unjustified absences to the tests.

IV – Research

1. General research into the genome

Portugal cooperates in the Human Genome Progam as far as our technical and economic capacities permit, namely through the Institute Ricardo Jorge.

Some clinical centres, mentioned in this report, are developing specific programs for the identification of certain diseases and the location of their origin. I would mention Haemoglobinopathia, Haemophilia, Cystic Fibrosis, Familial Amyloidotic Polyneuropathy (FAP), Down Syndrome, Phenylketonuria and Hypothyrodism and the Pathologies of sex determination (relevant for the legal problems of transsexuality). It seems clear that the greatest advances are in the study of FAP, whose mutation and place of origin are now precisely defined.

Of more concern to us here is however the fact that all research centres act according to firm deontological rules. The collaboration of the researched is seeked through very detailed information on the aims of the intervention, on the non-therapeutic character of the research; and no medical act is performed without free consent of the individual.

On the other hand, as screening and other testing are under Medical Genetics Jacinto Magalhães, in Oporto, the blood of a million Portuguese, meaning 10% of the population, is now in store.

Besides the services dedicated to the study of genetic diseases, also the Institutes of Legal Medicine keep the results of their activity (in the ambit of paternity suits and in the criminal domain) for the specific aim of establishing genic frequencies of the population.

In connection to this we might now refer the work on genetic therapy in relation to FAP.

The location of the gene is known, and also known is the constitution of the mutation that causes the production, by the liver, of a protein that concentrates in the plasma and in the muscles, beginning in the peripheral ones. Specialists think it is difficult to inhibit the production of the noxious substance but it seems certain that a decisive result will be obtained within a few years.

The development of this research has never raised any ethical or juridical problems. And when we reach the phase where it will be necessary to apply genetic therapeutics on a pacient no difficulty of the kind is foreseeable since no therapeutic attempt can be worse than the normal evolution of the disease; the strict respect for the general norms of informed consent should suffice.

Some difficulties may arise in the selection of candidates if the therapeutics can not be offered at once to all.

2. Estabishing of family banks

The most clear and interesting case of organization of family banks is the one related to FAP. About five hundred and sixty genealogical families are registered and this permits the identification of the population presently at risk.

At the Institute Ricaldo Iorge, in Lisbon, the state of families in relation to other genetic diseases with less expression in Portugal (Haemoglobinopathia, Haemophilia and Cystic Fibrosis) is also registered. For the former, including Thalassaemia and Sickle-cell anaemia, the register started with a national program based on a screening made during medical military inspections. Through this screening it was possible to identify the regions in the country deserving special attention because of a greater concentration of cases.

It has not yet been possible for me to know the exact terms of the information given to the screened population.

But it is important to conclude that, since the register is nominal, it creates serious responsibilities concerning the confidentiality of the data.

There are no specific national rules on the conservation of registers or on right of access to the information they contain. The people in charge however are perfectly aware of the gravity, for the people concerned, of any breach of secrecy. Access to the data is only allowed, therefore, to the persons concerned or with their permission. as is usually the case with the attending physician.

No one in charge has any knowledge of any request for information from employers or insurers. And all would reject any inforrnation for such purposes.

Access to the files is even denied to other physicians for scientific or academic purposes.

3. Ethical committees

As far as we know problems of human genome analysis have not been dealt with by "Hospital ethical committees" or by the recently created National Council for Ethics and the Sciences of Life.

Soon, however, the ethical commitee of Hospital Santo António, in Oporto, will be requested to appreciate a project on a therapeutic essay for FAP.