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A proposed organizational scheme for a research unit for the diagnosis and prophylaxis of genetic diseases

by

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An organizational scheme aimed at the prophylaxis and diagnosis of the hereditary and chromosomal diseases would provide for:

a) preliminary field-work; *b*) pre-conception out-patient clinics; *c*) pre-natal out-patient clinics.

As regards field-work, the primary phase would consist in promotional and screening work by the various health workers (physicians, obstetricians, social workers, health workers, etc.) in the day-clinics of the district, the health centres, and in the various places where people congregate (schools, housing estates, factories, etc.).

The second phase would provide for a comprehensive system of surgery assistance for couples who require genetic advice so that they can consider responsible parenthood, when impelled to seek such advice because of the actual or presumed presence either in a previous child or in a relative, or in the couple's own heredity, of an hereditary or chromosomal disease. They might also seek advice because of a right and legitimate desire for information.

The whole of the work to be carried out in the surgeries can be divided into

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two separate phases, pre-conceptual and pre-natal, which we shall examine individually.

A) PRE-CONCEPTUAL PHASE

This is the traditional way of dealing with genetic consultation, its ultimate aim being to prevent the birth of infants affected by genetic diseases, by calculation of the statistical probability with regard to the specific risk of any possible pregnancy. The calculation of such a risk can be made available (Fig. 1) from an

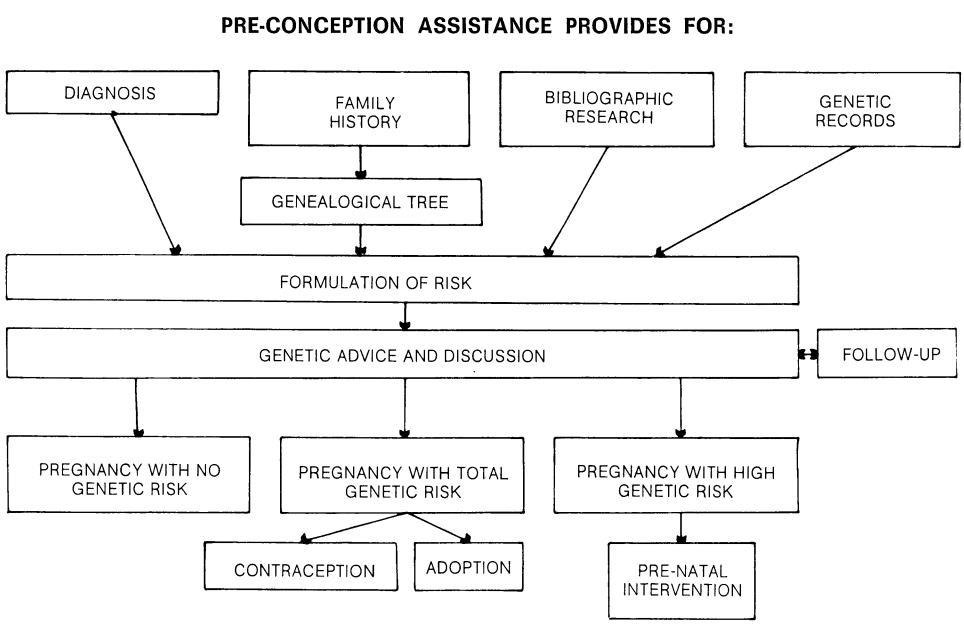

FIG. 1 - Organizational scheme for a research unit for the diagnosis and prophylaxis of hereditary and chromosomal diseases.

exact diagnosis of the disease, from the construction of a genealogical tree (being the graphic expression of a detailed family history), from the discovery of healthy carriers of recessive autosomal diseases, as well as from bibliographical research into the disease in question and from information inferred from the central genetic records. Once the possible risk has thus been formulated, it will be communicated to the patients and can be discussed with them both so that they can understand clearly and exactly the nature of the genetic advice.

Couples who have been assessed as running a « high genetic risk » and who, voluntarily or unwillingly, have undertaken a pregnancy, will be assigned to the second, or pre-natal, phase of surgery treatment.

B) PRE-NATAL PHASE

Genetic consultation has taken a great step forwards, qualitatively speaking, with the introduction of this type of intervention. It means that the parents have been offered an opportunity to understand the conditions of the conception. This fact,

apart from being their own incontestable right, provides them with basic information they need in order to make a conscious decision concerning the opportuneness of continuing with the pregnancy or not. The value of the pre-natal discussion is in any case not exhausted once genetic disease has been diagnosed; it is indicated also if there is any possibility of introducing effective therapy from birth onwards, and (in the future, certainly) even during intra-uterine life. Finally, as regards the purely speculative aspect, which is an indispensable factor before any clinical applications can be made, it is relevant to investigate the physiopathology of the foetus, which thus attains the status of a potential patient in its own right.

The techniques at present available to pre-natal investigation (Fig. 2) are ultrasonics and amniocentesis. Within a brief space of time, these techniques have facilitated the early diagnosis *in utero* of numerous hereditary and chromosomal diseases.

To sum up, we may thus imagine our organizational scheme (Fig. 3) as being a « *research unit for the diagnosis and prophylaxis of the genetic diseases* ». Considering that the point of departure consists of the birth of a child affected by possible genetic disease, or considered to be so, or of the existence of a couple of « high genetic risk », the first task will be to diagnose the disease by means of whatever instruments are part of the equipment of the centre (family planning investigations, etc.), while to some extent they must be composed of normal routine methods, aimed at discovering morbid conditions by means of mass screening. The findings thus obtained, the bibliographical research and the information

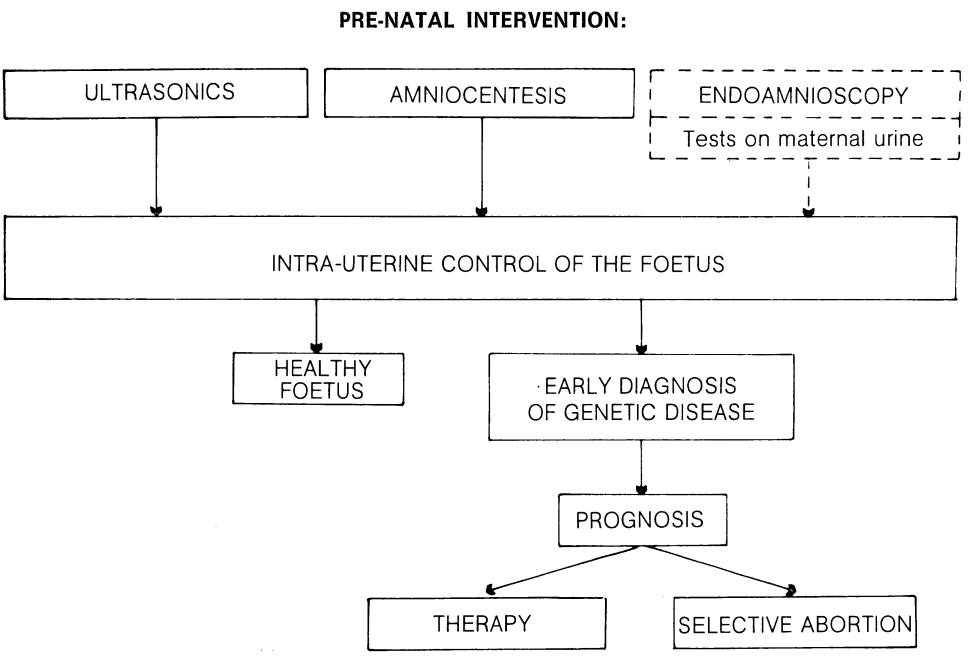

FIG. 2 - Organizational scheme for a research unit for the diagnosis of prophylaxis of hereditary and chromosomal diseases.

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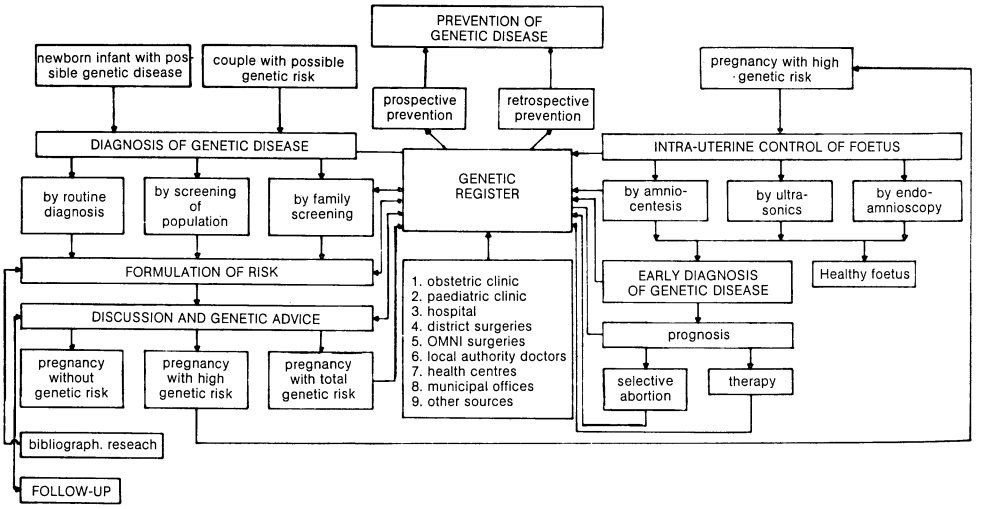

FIG. 3 - Organization scheme for a research unit for the diagnosis and prophylaxis of hereditary and chromosomal diseases.

derived from the central records will enable the risk to be formulated, and genetic advice to be given. At present, pregnancies that carry « no genetic risk » (the risk being no greater than is foreseeable for any pregnancy) leave the circuit which involves the genetic centre; those listed as « total risk » follow a shorter circuit, concerned with the prevention of the birth of an affected child, whose characteristics are supplied to the central registry; and finally, the pregnancies of « high genetic risk » are dealt with by second-stage intervention, involving pre-natal checks by the methods we have already illustrated. The early diagnosis *in utero* of « genetic diseases » will in its turn lead, whenever possible and convenient, to adequate therapy; where this is not possible, the prevention of the birth of a diseased foetus can only be brought about by genetic abortion.

Serum and placenta levels of cholestasis enzymes in pregnancy

by

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Various methods are presently available to diagnose intra and extrahepatic cholestastasis. Chemical analyses used mainly involve bilirubin and cholesterol levels

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