The Value Of A Genetics Counselling Clinic

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"Is it safe for me to have another baby, or will the next one be just the same as the last?" These words are heard over and over by doctors who have patients unfortunate enough to be delivered of babies deformed, physically or mentally, or suffering from a chronic illness. It is not good enough these days for the doctor to give either an emphatic uninformed "no" or a breezy go-ahead signal when expert advice on the subject is readily available. Parents are surely entitled to be advised of the latest research and to benefit in some way from it.

The science of genetics which started only about a century ago in its present concepts is at the moment the most rapidly expanding field of medicine, and almost every medical journal at the present time is found to contain at least one article on the subject. The fields of cancer cytology and virology are also very closely linked with genetics, and some of the most fascinating research can hardly be pigeon-holed into any of these separate fields.

Many people, including doctors, have an antipathy towards the genetic counselling clinic as it is synonymous in their minds with a birth control clinic, but this conception is entirely wrong. Not only is no birth control information at all available at a genetic counselling clinic, but no attempt is made to persuade persons to limit their family. Indeed in about half to one third of the cases, one is able to reassure people that they are in no way likely to produce defective children. The information available is objectively given and what use the persons make of it is entirely dictated by their own inclinations and principles. Most of the patients referred to a genetic counselling clinic are people who ask for referral. Many write for advice, perhaps prompted by reading articles in lay journals. Many others are referred by their own doctors who are uncertain of the exact mechanism of inheritance of certain diseases. Others are persons who wish to make enquiries about inheritance of diseases before they become married, and still others have queries about prospective adoptions.

In most cases a case history and family history of the patient are taken. These are taken in as complete detail as possible, with verification of pathological data by letters to hospitals etc. It is from these family histories and verified conditions that one's statistics of inheritance are compiled, and from which one can give likelihood of recurrence of the less well documented diseases. From studies of family histories and consanguineous marriages the preliminary suspicion that certain conditions are hereditarily transmitted may be obtained, especially when the diseases are rare and obviously not transmitted by dominant inheritance.

To give an example of a typical case referred to the clinic we might consider some of the more recently discovered enzyme defect diseases. These are usually inherited in a recessive manner, that is that two apparently normal parents (who may often be distant relatives of each other) produce a child who carries a double dose of the recessive gene which each of them has been carrying asymptomatically in single dose. Among these diseases are two causing severe effects in the young infant. I refer to phenylketonuria and galactosemia.

Phenylketonuria is a condition in which phenylalanine, which is present in quite considerable quantities in ordinary cow's milk and other common articles of diet, is not completely metabolised by the defective individual due to an enzyme defect in the intermediate breakdown of phenylalanine to tyrosine. The presence of large quantities of the intermediate by-product is thought to have a deleterious effect on the brain and produces, among other defects, severe mental retardation. When a

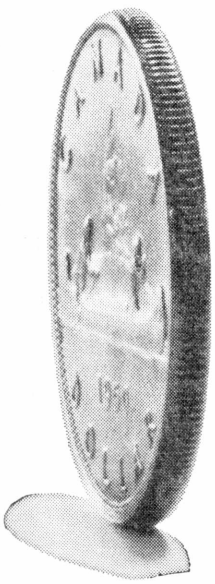
pilot case has been discovered in a family, although it may be too late to give maximum benefit to the first unfortunate child, a great deal of benefit for the whole family can result from our genetic knowledge applied in conjunction with the metabolic department. First of all in any future pregnancy, almost as soon as the child is born, tests can be performed to ascertain whether the new child will be affected or not. If this is found out within the first few weeks (it sometimes takes this time for the phenylketones to be present in the urine), a special diet low in phenylalanine can be substituted for the usual formula. Statistics show that the future of the early treated children is very satisfactory, but this is not all. Tests on the relatives of the affected child and his parents can be done to discover whether they are asymptomatic carriers of the gene and advice given for future marriage. This would include assuring many of the relatives that they do not appear to be carriers of the defect.

Galactosemia, although a different disease entirely, and causing entirely different symptoms, acts in exactly the same way. Again tests are available to find new cases early and soon it will be possible to test the asymptomatic gene carriers by an estimation of the enzyme carrying properties of their erythrocytes. Treatment for prevention of ill effect is available in special diets for the newborn and affected infants. You may be interested to know that a study is in progress at present in cooperation with the Grace Maternity Hospital, the Halifax Infirmary, the Sydney City and St. Rita's Hospitals, to discover the incidence in this area of phenylketonuria and other similar diseases caused by enzyme defects. Mothers of newborn babies are given small discs of filter paper which they take home. At approximately 6 weeks of age they allow the baby to urinate on the paper and dry it off. The dried paper is sent to the Research Laboratory at the Children's Hospital, Halifax, where it is tested for the presence of phenylketones, as well as acid mucopolysaccharides (Hurler's syndrome), maple syrup urine disease, galactose, fructose, sucrose and glucose. To date approximately 750 papers have been sent into the lab and one case of galactosaemia encountered. There have been eight false positives on which a second specimen has been requested and these have been normal. This is an interesting and informative study as 1% of institutionalized mental defective persons in England are said to suffer from phenylketonuria, and it is interesting to discover whether there is the same percentage in this country or not.

Other problems frequently dealt with at a genetics counselling clinic are the premarital worries of the person in whom a family disease is present. Such diseases can be neurofibromatosis, hemophilia, Huntington's chorea, epilepsy, hare lip and cleft palate, etc. Whereas the hereditary mechanism of neurofibromatosis is well known, that of hare lip and cleft palate is by no means clear and there are several different factors to be considered here. Many hare lips and cleft palates are not hereditarily transmitted at all, and a careful investigation of all the factors involved is required for a considered opinion to be given to the parents. In the case of hemophilia, which we all know is a sex linked recessively transmitted disease, the abnormal gene is handed by the unaffected female to some of her male offspring, while some of her daughters will themselves be asymptomatic carriers. The family wants advice on marriageability of their members. Apart from the inadvisability of choosing his female cousin as a spouse, the unaffected male has free choice with no worries. However, half the sisters will be carriers. Some of the female carriers appear to have a partial deficiency of anti-hemophilic globulin in their blood which may be detected, and of course these female carriers will also have the risk, no matter whom they marry, of producing hemophilic sons and carrier daughters. However, tests are not far enough advanced at the moment to be quite sure of the certainty of this method, and whether it will be possible to detect those who are free of the disease by such tests. It would be a great relief to those who know the suffering of living in a family of hemophilics, if we were able to pronounce the normal daughters of a hemophilia carrying mother as normal.

In the queries by prospective adopting parents the clinic is in a much more difficult position. Whereas adopting parents are entitled to full health information on the child they are adopting, the information available from the very nature of the birth, makes family history of both natural parents almost impossible to obtain. Sometimes the adopting parents approach the genetics counsellor to ask him to investigate the child they propose to adopt. The counsellor usually approaches the doctor who examines the child for adoption and the society social worker who is arranging for the adoption for information. Although the grosser errors may be avoided, only in cases where there is a dominant disease in the natural parents would the investigation be of any certainty, and this has to be pointed out to the adopting parents in language they can understand.

Most of the teaching hospitals in Canada have an attached genetics counselling clinic, and in Halifax we are beginning to offer the service also. Fortunately most most of the workers of these clinics have liaison with the other clinics and are able to pool their knowledge, so that the widest use of what knowledge is available can be made.



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