Some Aspects of Medical Genetics in Island Populations

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The study of island populations is of considerable potential interest for the investigation of genetic problems. There may be an increase in consanguineous marriages but popular guesses as to the extent to which breeding groups on an island are closed are often wrong, and this can be checked against parish register material. Furthermore, as was found in Japan, inbreeding may not necessarily be deleterious. The pattern of congenital malformations may be different from that of the mainland and a register of these should be kept, as among other things this is a useful stimulator not only of further research but also of better social care. Again, because of genetic drift, whereby chance exercises a considerable influence on gene frequencies, blood group distributions may be unusual and unfavourable mutations persist in isolated communities. These factors will be discussed in relation to the Isle of Man.

Genetics Survey of the Manx

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In recent years physical anthropologists and human biologists have carried out numerous surveys of the genetic characteristics of populations. In such a context this paper is a preliminary report on a study of genetic variability in the Manx population. Included in the survey were those individuals having three or four grandparents born on the Isle of Man. Results are given for the following unifactorial traits: most of the blood group antigenic systems, including ABO, Rhesus, Kell, Duffy, Lu, and Kp, the red blood cell isoenzyme acid phosphatase, and the plasma proteins haptoglobin and transferrin. Non-serological inherited factors such as phenylthiocarbamide testing are also reported upon.

For some of the serological factors comparisons were made between blood donor and non-blood donor samples. Moreover the incidence of the genetic features was compared with those found in contiguous areas of Britain, particularly south west Scotland and Cumberland.

Mention is made of previous anthropological studies of the Island's population and their main findings. All previous work was of an anthropometric rather than serological nature. One aim of the present work is to discover whether genetic factors complement the variability in the Manx population found in the previous studies.

A very brief account of the main periods occurring in the Island's history is given because of its possible use in helping to interpret the results of the present survey.

Erythroblastosis Foetalis

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Erythroblastosis foetalis (haemolytic disease of the newborn) is an important, if decreasing cause, of perinatal death. Results obtained during the perinatal mortality survey (1958) (Claireaux, 1963) showed that it was responsible for 4-0% of all perinatal deaths and the incidence was 1-4 per 1,000 live and stillbirths. The final results of a similar study, British births (1970), are not yet available but preliminary evaluation of postmortem material shows that it is still a factor in perinatal death but the incidence is probably lower than in 1958.

Erythroblastosis foetalis was a term coined in 1933 to include the three main types of the disease: (a) hydrops foetalis; (b) icterus gravis neonaetorum; (c) congenital haemolytic anaemia. This last is not to be confused with congenital spheroctysis. Hydrops foetalis was the most severe form of the disease and usually resulted in a stillbirth or in the delivery of a moribund infant. Icterus gravis was, if untreated, also likely to have a fatal termination and the infant succumbed as a result of brain damage (kerinaetosis). The remaining cases, much less severe and seldom fatal, were clinically regarded as examples of haemolytic anaemia. The clinical features of the disease and the pathological changes in fatal cases were known some considerable time before the discovery of the Rhusov blood groups (1940). It was only then that it became clear the condition resulted from iso-immunization of the mother by a blood group she did not possess.

Reference


The Role of RH Antibodies in Causing Haemolytic Disease of the Newborn and in Preventing It

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HAEMOLYTIC DISEASE OF THE NEWBORN

The relation between the amount of anti-Rh on the red cells of an Rh-positive infant and the severity of the haemolytic process is not very close; one reason may be the varying ability of different examples of anti-Rh to bring about red cell destruction.

In haemolytic disease of the newborn the concentration of anti-Rh in the plasma of the Rh-positive infant is always very much lower than that in the mother's plasma, emphasizing that the relatively slow transfer of IgG across the placenta plays a crucial role in mitigating the severity of the damage. Because of this slow transfer, it is quite safe to give injections of the order of 200 μg anti-Rh to Rh-negative women who are pregnant with an Rh-positive foetus. Incidentally, even if such a dose is accidentally injected into a newborn Rh-positive infant, it causes only very mild haemolytic disease.

PRIMARY RH IMMUNIZATION

A single injection of Rh-positive red cells induces primary immunization in about 65% of Rh-negative subjects. With a relatively large dose (200 ml cells), anti-Rh is as a rule readily detectable three to six