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BIBLIOGRAPHY

- Brown, H. C.: *Lancet*, 1928, i, 388.
 Brown, H. C., and Davis, L. J.: *Brit. Journ. Exper. Path.*, 1927, viii, 397.
 Brüssin, A. M.: *Zeit. f. Immunitäts.*, 1925, xlv, 328.
 Fairley, N. Hamilton: *British Medical Journal*, 1934, ii, 10.
 Fletcher, W.: *Trans. Roy. Soc. Trop. Med. and Hygiene*, 1927, xxi, 265.
 Kritschewski, J. L., and Tschirikower, R. S.: *Zeit. f. Immunitäts.*, 1926, xlv, 530.
 Laveran A., and Mesnil, F.: *Ann. de l'Inst. Pasteur*, 1901, xv, 673.
 Postmus, S.: *Nederl. Tijdschr. v. Geneesk.*, 1933, iv, 2648.
 Schüffner, W.: *Trans. Roy. Soc. Trop. Med. and Hygiene*, 1934, xxviii, 7.

HEREDITY AND MENTAL DEFICIENCY*

BY

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Those members of the medical profession who are not familiar with the science of genetics and its application to human beings may not unreasonably ask why it is not sufficient to collect a large number of pedigrees of mental defectives, and, having analysed them, settle the problem of heredity and mental deficiency once and for all. This paper attempts to explain why this simple procedure does not, and cannot, provide all the necessary information.

Diversity of Views Held To-day

In spite of the numerous researches carried out on this subject, very divergent views are held by different authorities. For example, Professor McNeill¹ recently published a paper in the *British Medical Journal* entitled "Heredity a Minor Factor in Mental Deficiency." As a contrast, a passage may be quoted from the article on mental deficiency by Dr. Rudolf Pintner in the *Handbook of Child Psychology*.² "The great majority of feeble-minded individuals are simply born that way. They occur in families where feeble-mindedness or neuropathic conditions are common. No causal factor can be discovered. There may be secondary or aggravating factors, but not sufficient to cause feeble-mindedness. We therefore say that feeble-mindedness is inherited. The percentage of such inherited cases is put at about 90 by Hollingworth and about 80 by Tredgold. Amentia caused by disease or accident is therefore a relatively minor consideration.

The complete and final proof of a hereditary mechanism is the demonstration of Mendelian ratios. When inheritance is simple and uncomplicated, and when non-genetic influences are not operating effectively, this is an easy task experimentally, and not too difficult in the human subject. If mental deficiency were as simple as this the whole subject would have been settled long ago. Clearly, complexities exist that make genetical analysis much more difficult.

Mental Defectives Cannot be Sharply Separated from the Rest of the Population

The first difficulty is that mental defectives do not form a class that can be sharply distinguished from the rest of the population. The legal criterion is essentially one of

social adaptation. Where precisely that line is drawn does not have any biological meaning. In terms of the closely related, but not identical, intelligence tests, the distribution of mentality is continuous. Where the line is drawn that separates normality from mental deficiency is arbitrary. Whatever criteria are adopted, therefore, a sharp line of division cannot be secured. Dullness and retardation shade off into mental deficiency.

This does not mean that in certain cases mental deficiency cannot be due to a simple genetic difference. The continuity of the distribution may be partly due to the way that intelligence is measured. It is more likely, however, that many defectives, especially those falling into the legally feeble-minded group, do represent the lower end of the ordinary curve of variability, just as individuals occur who differ from their fellows in being very short.

Lewis³ has elaborated this conception, and terms those to whom it applies the "subcultural" group. This group is distinguished from those defectives in whom the mental state is the result of a definitely pathological condition, hereditary or otherwise. If, then, many defectives are to be regarded as extreme variants at the lower end of the curve of distribution of intelligence, it is clear that biometrical researches are required. Large numbers of individuals must be measured, and, in addition, it is not sufficient merely to contrast defectives and normals. Quantitative measurements are necessary, and the problem must be approached in the first instance by studying mentality in general—that is, all grades in the population. Biometrical studies on relatives are capable of yielding much information as to the part played by heredity in determining any characteristics that can be measured.

Mental Deficiency is not a Clinical or Genetic Entity

Another complication is due to the fact that the term covers a number of conditions, that may or may not be clearly distinguishable. Mental defectives have in common an underdeveloped and an ill-developed nervous system, but they show among themselves many differences both of kind and of degree in this state of underdevelopment. These differences are also found in mental defectives in regard to physical characteristics such as stature or size of head. Mental deficiency, then, is neither a clinical nor a genetic entity. It is not a genetic entity because it is found that there are genetic differences between groups selected in different ways. For example, it is generally accepted that those showing gross defect have fewer affected relatives than those with lesser degrees of abnormality. Mongols, too, differ from other defectives in having fewer affected relatives, and in addition the occurrence of mongolism shows a clear association with advanced maternal age.

Clinical and Genetic Entities are not Identical

It is important to recognize that clinical and genetic entities often do not correspond. For example, congenital stationary night-blindness may be inherited as a Mendelian dominant, as a recessive, or may be sex-linked. Clearly, therefore, at least three, and probably more, genetic factors exist, any one of which is capable of producing the same clinical end-result. It can be shown in other cases, such as albinism, that any one of several factors that show the same method of transmission may be responsible for the condition. Sometimes there are slight differences, constant for the family group, that indicate that different factors are at work in different cases. In two families that I had the opportunity of observing there were a number of members suffering from congenital cataract of the same type, and in both

* The substance of a paper read to the Maternity and Child Welfare Section of the Society of Medical Officers of Health, November 30th, 1934.

families inherited as a dominant. In one family, however, the age of onset was much less and the condition progressed much more rapidly. It is likely, however, that in addition to instances in which slight clinical differences indicate a genetic difference, there are many instances in which there are no corresponding clinical differences.

The Interaction of Genetic Factors, and the Interaction of Genetic and Non-genetic Factors

Genetic factors are often associated together in producing the end-result. For example, a comparatively rare gene may produce a condition, but there may also exist a relatively common gene (or genes) that can prevent the first factor from producing its effect. Similarly, non-genetic influences may inhibit the action of a genetic factor. In yet other instances other genetic or non-genetic factors may reinforce the action of the main gene. Instances of interaction of this kind may be of any degree of complexity. Ultimately we get multifactorial inheritance. This has already been mentioned as a likely mechanism in many cases of mental deficiency. It will usually be found that environmental, or, more strictly, non-genetic, influences are also at work. Perhaps the most usual case would be to find that the end-result is due to the action of unfavourable environmental influences acting upon the basis of a poor hereditary potentiality.

A Single Genetic Factor may have Diverse Clinical Results

If a genetic factor (or factors) is the essential condition for the appearance of a defect, and if its action can be modified in the way just described, it is to be expected that one and the same essential genetic mechanism may yield quite different end-results. Osler's disease (multiple telangiectasia) is very regularly inherited as a Mendelian dominant. In a typical case the characteristic angiomatous areas are seen, and the patient suffers from repeated epistaxis. Relatives, however, who undoubtedly possess the same factor may show few angiomas or even none, though they suffer from epistaxis. In other instances the only symptoms or signs are haemoptysis or haematuria. Progressive pseudo-hypertrophic polyneuritis is also a Mendelian dominant, but some relatives who possess the factor may have only absent deep reflexes or pes cavus.

In relation to mental deficiency it is impossible to deny the importance of what is termed "neuropathic diathesis." In one and the same family group are found mental defect, insanity, epilepsy, chronic alcoholism, and so forth. It is difficult to avoid the conclusion that there is an underlying weakness of the nervous system that is common to members of the family group, but that the particular manifestation depends on other things, either subsidiary genetic factors or non-genetic factors, or both.

Mental Deficiency as a Biological Problem

In the British Medical Association's report on mental deficiency⁴ it was very properly pointed out that mental deficiency is as much a biological as a social problem.

To the biologist, then, reviewing the subject of mental deficiency with a view to research on its relation to heredity, the problem appears as follows. Mental defectives can be regarded either as the extreme minus end of the curve of distribution of intelligence or as the result of a definite abnormality in development, genetic or otherwise. The first conception applies more especially to high-grade cases and the second to low-grade cases. Amongst recognized defectives are to be found clinical differences, some fairly clear-cut, others allowing only a very partial separation into groups. Genetically we shall

expect to find various types of inheritance, both simple and complex, heredity interacting with environment, and, finally, non-genetic cases. To complete a complex picture, while gross cases are readily distinguishable as abnormal, there is no biological line that separates the high-grade defective from the dull normal.

Perhaps it is only fair to say that some observers might maintain that the picture is simpler than this. They would hold that a relatively simple hereditary basis exists rather on the lines mentioned in connexion with neuropathic diathesis, and that it is largely due to imperfection of technique that defectives and normals cannot be simply contrasted. The essential underlying hereditary mechanism might be simple and the same in very many cases, and the apparent diversity grafted on afterwards. To express a personal opinion, I consider such a view much less likely to be true than the picture I have presented. Whether the complexity is real, as I think most probable, or largely apparent, much the same difficulties have to be faced, and the data must be collected in the same way.

Inadequacy of Pedigrees

It might still be asked why straightforward pedigree collection cannot provide much of the solution. The answer is that a single pedigree by itself cannot give a clear-cut answer except in very simple cases. A simple Mendelian dominant can often be recognized on the basis of a single large pedigree, and so can sex-linked inheritance. Recessive inheritance can rarely be deduced from a single pedigree, and then only with some degree of likelihood. In the same way the introduction of any complexities makes it almost impossible to secure clear-cut results from a single family. In one form or another families must be added together, but, unfortunately, in the case of mental deficiency we do not know what we are adding together.

Even in the most favourable case—the simple dominant—there are difficulties. Mental defect cannot usually be inferred with certainty about people who are dead. It is also necessary, if this simple hypothesis is to be tested, to know whether the "normals" in the family were really normal—that is, distributed something like the general population—or whether they were, on the average, mentally retarded. This last finding would make the simple dominant hypothesis very doubtful. It is difficult enough to classify adults even if they are seen, and practically impossible if they are not. The clear-cut and obvious physical deformity may lend itself to a greatly extended pedigree, and details of previous generations may be sufficiently accurate, but in mental deficiency the difficulties are far greater.

The Correct Approach

Much has undoubtedly been learnt in the past from the study of individual pedigrees, but greater advances may confidently be expected from the application of modern methods based on the integration of biometry and genetics. A line of investigation that is now especially required is the study of the mentality of a complete cross-section of the population. The biometrical ideal, a random sample, is very difficult to attain. A more practical plan is to select a suitable area and to investigate, as far as possible, all the children of certain ages living within it at a given date. It is to this aspect of the problem that the main activities of the Burden Mental Research Trust are now being directed.

Whilst this paper has been chiefly concerned with the limitations of pedigree collection, the constructive side of the problem has not been forgotten, and it is hoped to discuss this more fully on a future occasion.

Summary

It is sometimes asked why it is not sufficient to collect a number of pedigrees of mental defectives, and, having analysed them, settle the problem of heredity and mental deficiency once and for all. Both from the clinical and from the biological point of view the problem is a complex one, and a number of considerations are discussed that make it necessary to adopt more complicated lines of approach.

REFERENCES

- ¹ McNeill, C.: *British Medical Journal*, 1934, i, 584.
- ² Pintner, R.: *A Handbook of Child Psychology*, Clark University Press, 1931, p. 615.
- ³ Lewis, E. O.: *Journ. Ment. Sci.*, 1933, lxxix, 298.
- ⁴ British Medical Association, Report of the Mental Deficiency Committee, July, 1932.

REPAIR OF PERINEAL TEARS

A SIMPLE TECHNIQUE

BY

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Happily, extensive tears of the perineum are seen very infrequently nowadays. Tears involving the vaginal wall are still found in such cases as a primipara with a short quick labour, and in a multipara with much scar tissue, the result of a former tear, at least as often as in cases where forceps have been applied. The rates of maternal mortality are directly influenced by imperfectly repaired tears of the perineum. Sepsis is likely to develop when a tear is stitched in such a manner as to leave pockets and tags, or when a high vaginal or cervical tear is overlooked altogether. The general practitioner need never hesitate to investigate thoroughly, and feel satisfied that he has performed an efficient repair of any tear found if the following simple technique is carried out in all cases.

The Method in Detail

When sterilizing the forceps (whether to be used subsequently or not) there should be included in the sterilizer three rubber gloves, two artery forceps or needle-holders, two large half-curved needles, two small half-curved needles, a few strands of silkworm suture, scissors, a double speculum, and a flat-bladed tongue forceps or a vulsellum. Tubes of sterilized catgut should also be available.

Immediately after the birth and separation of the child (and while the mother is still presumably under anaesthesia) the patient is placed on the left side, with the buttocks over the raised edge of the bed or over a small pillow placed under the accouchement sheet, so that the drawn-up knees are on a lower plane than the buttocks. This position will permit the uterine discharges to flow forwards, downwards, and away from the area to be dealt with. A good light is essential.

The third rubber glove is pulled over the already gloved left hand. The left index finger is gently inserted to its full extent into the rectum. This finger is then swept forwards and downwards through an arc of 90 degrees, bringing practically the whole length of the posterior vaginal wall into the vaginal orifice, and so into full view.

If a tear in the vaginal wall be discovered, this is dealt with by a small needle and a continuous catgut suture, beginning at the uppermost limit of the tear. The edges should be carefully approximated by stitches no more than 1/4 in. apart. The second finger and thumb of the left hand can be utilized to help to tie knots and to retain the strand of catgut while the stitches are applied by the right hand. This suture is tied off on

reaching the lower limit of the tear or on reaching the perineum if the tear extends into that part.

Should the tear be a very deep one, involving the sphincter and part of the rectum, the rectal tear is first repaired by interrupted catgut sutures from above downwards, with the knots tied off in the rectal cavity. The finger is then inserted in the rectum, as described, and a few interrupted catgut sutures applied from above downwards to bring the deeper tissues together. The sphincter is dealt with by applying a catgut suture deeply from one side to the other, taking in the anterior third of the circumference of the anal orifice. The vaginal aspect of the tear is now attended to, as previously described. Elongated tags, such as those caused by bifurcations of the tear, should be snipped off rather than run the risk of a slough later on.

When the perineum remains to be dealt with, or when only the perineum is involved in the tear, the two distal phalanges of the left index finger in the rectum will be found sufficient to make the perineal wound bulge downwards, exposing it in its full extent. A large needle, with silkworm suture, is applied in a deep stitch, taking in the perineal body. The finger in the rectum permits the needle to make a deep and sweeping course from one side to the other without fear of penetrating vagina or rectum. Two such sutures are usually sufficient in tears of the complete perineum. Before tying these sutures the finger is removed from the rectum, the outer glove stripped off the left hand, and the gloved hands swilled in antiseptic solution. The perineal sutures should not be tied tightly. One or two additional interrupted shallow or skin sutures may then be put in to exactly approximate any gaping skin edges of the perineal tear. A loose strip or tag should be snipped off.

After the placenta is expelled (I have never seen a placenta, in its passage down the vagina, break down a repaired tear of vagina or perineum) and the uterus is firmly contracted, the cervix should be examined. With the patient in the same position a speculum is gently inserted along the (repaired or intact) posterior vaginal wall and the cervix viewed by the light of a torch held by the nurse. If a cervical tear be discovered the nurse is asked to kneel on the bed behind the patient's back, and, only if it be well contracted, to press the fundus of the uterus gently down into the pelvis. With this manoeuvre it is surprising how near the vaginal orifice the cervix will present itself. A flat-bladed tongue forceps, in preference to a vulsellum, is made to take a light grip of the cervix, and may be safely handed to the nurse, to be held to one side. The cervical tear is repaired, with a small needle in holder, by catgut suture, beginning distally to the cervical opening.

It is important after this operation to see that the uterus returns to its normal position. This should, owing to the possibility of a retroversion, be confirmed in all confinements by pressing the edge of the right hand deep into the abdominal wall, between the pubes and the body of the uterus, sweeping the latter upwards if necessary.

Anaesthesia is obtained by chloroform in a Junker's outfit, which is tied to the head of the bed or otherwise suspended in full view of the operator and beyond the patient's reach. The mask and bellows are manipulated by the patient, with help from the nurse, under the operator's instructions. The nurse should wear rubber gloves for ease in cleansing, and face masks are worn.

Advantages of the Method

The advantages of applying this technique in all cases are as follows. (1) Its simplicity and possibility of being carried out without assistance other than that of a nurse. (2) No tears are ever missed. (3) Hidden areas are easily