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HEALTH BULLETIN for TEACHERS

The Physical Basis of Heredity

The transmission of hereditary characteristics from one generation to the next naturally could not be explained until the respective roles played by each parent in the formation of offspring had been determined. The discovery of the male and female germ cells, or gametes, was not made until the 17th century. The female germ cells, or ova, were discovered by Reinier de Graaf and the male germ cells, or spermatozoa, by Johann Ham, a pupil of Van Leeuwenhoek, the Dutch microscopist. These discoveries caused a tremendous sensation. Most biologists of the day separated into two camps - the Ovists and the Animalculists. Both upheld the preformation theory, namely that the germ cell contains a completely developed embryo - a tiny male or a tiny female - with all its limbs and organs formed. The Ovists believed that this homunculus (little man) existed in the ovum and that the mother was the sole procreator; the Animalculists believed it existed in the head of the spermatozoon and that the father alone reproduces himself in his offspring.

In 1875, Oskar Hertwig actually saw the entry of a spermatozoon into the ovum of a sea urchin. Until then no definite information had been obtained regarding the mechanism of fertilization. Hertwig's observation clearly meant that both male and female germ cells had a part to play in the formation of offspring. Now it is known that the starting point for the development of a new individual is the fusion of two cell nuclei - the nucleus of the sperm with the nucleus of the egg. Since the fertilized egg is the sole connecting link between two successive generations, it must contain potentially everything that can be passed on from parents to child.

What Are These Hereditary Factors? Cytologists - students of cell structure - have discovered that within the nucleus of every dividing plant and animal cell there are microscopically small ribbon-shaped structures, the chromosomes, whose number is constant for each species - 48 in man, 8 in the fruit fly, 20 in maize, 16 in the cat, 38 in the rat, and so on. Strung along each chromosome like beads on a string are the genes, solid particles in which the hereditary characters are believed to be localised. The chromosomes are arranged in the nucleus in pairs according to their shape. During cell division each chromosome splits in half lengthwise thus doubling the number. Half of each chromosome goes to each daughter cell so that the nuclei of the two daughter cells contain exactly the same number of chromosomes as that of the mother cell.

The Formation of Gametes. When gametes - ova and spermatozoa - are formed from germ cells, the chromosomes do not split in two in the last division. Each gamete receives only one half the total number of chromosomes in the parent germ cell, one from each pair. Thus the head of a human sperm contains not a miniature man, but 24 chromosomes, the contribution of the father to the physical heritage of his child, and the nucleus of the ovum contains 24 chromosomes, the contribution of the mother. During fertilisation the number of chromosomes characteristic of the human species is automatically restored. The fertilised ovum with its 48 chromosomes then produces a human infant, not by the symmetrical enlargement of a mythical homunculus but by cell division and cell specialisation largely controlled by the "genes."

Boy or Girl? Every cell of the body throughout life contains a replica of the 24 pairs of chromosomes with which life for the individual began. Twenty-three pairs are sexless; that is, any one pair might just as readily be in a man as in a woman. The 24th pair determines sex. The two female sex

m chromosomes in a pair (known as X-chromosomes) are exactly alike. In a pair of male sex chromosomes, on the other hand, one is similar to the two female X-chromosomes and the other, called the Y-chromosome, is much smaller. When ova are formed from female germ cells each ovum will cotain one X-chromosome. When spermatozoa are formed from male germ cells, some will contain one X-chromosome, and others will contain one Y-chromosome. During fertilization there are two possibilities. Either an X-chromosome sperm will fertilize an X-chromosome egg and the result will be a girl (XX); or a Y-chromosome sperm will fertilize an X-chromosome egg and the result will be a boy (XY). Thus sex is determined at the moment of fertilization and is simply a matter of chance.

Thomas Hunt Morgan, the world's outstanding geneticist, advanced and later proved the hypothesis that the hereditary factors - the genes - are present in the chromosomes. Morgan was singularly fortunate in his choice of the fruit fly (Drosophila melanogaster) as his experimental animal. In genetic experiments the experimental animal or plant must possess two characteristics - great fertility and a short reproductive period. The little flies that one sees hovering in swarms over overripe fruit fulfill these conditions admirably. Although genes cannot be made visible by any known method - probably they are single molecules and no one yet has seen a molecule - Morgan and his co-workers have succeeded in making chromosome "maps" on which the genes are localized at very definite points in each chromosome. At present the precise position of about 500 genes of Drosophila is known. These genes determine eye color and form, body color and shape, wing length, shape, and position, and many other characteristics.

The Mendelian Laws. The discoveries on which our knowledge of the physical basis of heredity is founded were antedated by the establishment of the basic laws of inheritance. The first scientist to work out an intelligent

method of studying the inheritance of family resemblances was the Augustinian monk, Gregor Johann Mendel (1822-1884). Mendel decided to confine his studies to the inheritance in garden peas of one trait at a time. In each instance he crossed two pure strains of garden peas which differed only in one particular trait and then recorded the results of the cross through several generations of self-pollinated descendants.

Mendel determined that the first generation of offspring of a cross all resemble one another as well as one of the parents. Thus, if a tall pea plant is crossed with a dwarf pea plant all the offspring are tall. On inbreeding the tall hybrids of the first filial generation, the short form reappears in the second generation in the proportion of one dwarf to three tall. The dwarf offspring produce dwarf descendants through all succeeding generations. One third of the tall plants likewise produce only tall plants in succeeding generations. The remaining two thirds of the talls propagate themselves in the proportion of one pure dwarf to three talls. Thus they are hybrid talls like the offspring of the original cross.

Mendel interpreted these results as meaning first that inherited characteristics are produced by hereditary factors (we call them genes) which are passed along unchanged from generation to generation (the law of unit characters). Second, genes occur in each individual in pairs consisting of two similar genes or two different genes. If the individual possesses two different genes in any given pair one dominates the other so that it may be called a "dominant" and the hidden or latent factor a "recessive" (the law of dominance). Thus hybrid offspring of a cross between tall and dwarf garden peas and all tall because the trait tallness is a "dominant" and the opposite trait dwarfness is a "recessive." Third, since the recessive trait appears in the next generation, the dominant and recessive traits in each pair of genes cannot unite or

bland but must segregate out when the hybrid forms its gametes with one of every two mated genes going to each gamete (the law of segregation). Thus, some gametes would contain only the dominant character (a) and others would contain only the recessive character (b). In regard to a particular trait, therefore, the individual formed by the union of any two gametes may possess two dominant (a) genes (aa), or two recessive b genes (bb), or one of each kind (ab). The ratio in which these combinations occur in the offspring of hybrids is one pure dominant (aa), to two hybrid dominants (2ab), to one pure recessive (bb), (the law of hybridization).

Mendel embodied the results of his nine years of research in a paper which he read before the local natural history society in the year 1865. This paper, subsequently published in the proceedings of the society, caused no stir in the scientific world and was promptly forgotten. Mendel himself seems not to have realised the significance of his work. He became the abbot of his monastery, dropped his scientific studies, and died 16 years before three botanists working independently of one another simultaneously rediscovered the Mendelian laws. In that year, 1900, modern genetics, the science of heredity, was born.

Human Genetics. Man's chromosomes and genes behave as do those in garden peas and fruit flies. But it is one thing to work out the laws of inheritance under experimental conditions and starting with pure strains of plants or animals; it is quite another to apply these laws to human heredity. The list of man's variable characters could be extended almost indefinitely. It is one of the tasks of the human geneticist to find out which of these variable characters have a hereditary basis. In other words just what characters can be inherited and what can not? Most geneticists no longer believe in the inheritance of acquired characteristics. The changes made in an indi-

vidual by what he does or thinks and what is done to him during his lifetime would have to be accompanied by corresponding changes in specific genes if any of his desirable or undesirable acquired characteristics were to be transmitted to his offspring. A tremendous amount of evidence has been accumulated to prove that this does not happen.

So far as the improvement of the race is concerned the question as to what human defects, diseases, and abnormalities have a hereditary basis is of the greatest importance. In the biological sense an imperfection is considered

hereditary only when genes are involved. Thus syphilis is not hereditary because the spirochete responsible has no effect on the genes but is merely transmitted from the mother to the developing child.

Most hereditary defects are controlled by recessive genes so that they show up only when each parent contributes to the fertilized ovum the recessive genes responsible. For example, a man and a woman may each carry one recessive gene for feeble-mindedness and be perfectly normal because the recessive gene is supressed by a dominant normal gene. But if these individuals should marry and have children there is one chance in four that a sperm carrying the gene for feeble-mindedness may fertilize an ovum carrying the same recessive gene. The result will be a feeble-minded child.

Heredity Versus Environment. The fact that genes ordinarily are passed on unchanged from generation to generation does not mean that they cannot be altered. When a changed gene occurs, a transformation or mutation of the characteristic which it influences is produced in the offspring. Changed or "mutated" genes are responsible for innumerable new strains in animals and plants. In 1927 H. G. Muller, a pupil of Morgan's, discovered that mutations in the fruit fly can be produced experimentally by bombarding them with X-rays. There is evidence also that heat, ultraviolation radiation, and certain chemicals also can induce changes in genes. Mutated genes apparently are produced by environmental factors of one kind or another and thus environment may be said to have the power of striking at the very roots of life.

However, it is not the effect of environment on the genes that concerns most people. The important question for the individual is, "which has the greater effect on me, my unchangeable inheritance or my changeable environment?" ~~The classic proponent of the theory that environment counts for little is Hans Christian Anderson's ugly duckling: "It matters nothing if one is born~~

~~in a duck-yard, if one has only lain in a swan's egg.~~ ~~Certainly the careers of many great men and women would bear out this theory. On the other hand who can tell how often the duck-yard has presented obstacles too great even for a swan?~~

Whatever may be the weight attached to inheritance or environment in influencing human destiny, it is certain that the individual is a product of the two and that the environment from the moment of conception is the factor most readily controllable. The Eugenists ~~(advocates of the science of being well born)~~ hold out hopes for an improved mankind in the selection, (through the regulation of human breeding) of the genes we pass on. However, recessive, defect-producing genes are too widely distributed in all human families to make possible the elimination of even obviously undesirable genes (such as those for feeble-mindedness) for many generations to come. As for the others, who can say ~~what genes are "good" and what are "bad"~~ until we have put all persons in environments that will enable them to make the best of what they have?

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