

CHAPTER II

GENETICS IN BIOLOGICAL PERSPECTIVE

Since long time ago, people knew from their daily experience that the child often had traits as like as their parents. Not only in physic, but also in their character and behavior. However, in this world no one is exactly same with others. All people have differences in inherit characteristics.

In daily life, we also find many terms about the resemblance of a child with her parents, such as 'fruit falls not far from the tree' and 'the stream of water falling into the cesspool too'. Both mean a child has similarities with both parents, in physical form, behavior, and his favorite. But not all the characters inherited to his son, there are certain criteria that must be completed when it characters could be called the character of heredity. This is where genetics is needed to determine the inherit characters of living thing especially human being.

A. DEFINITION AND ITS SCOPE

1) Definition of Genetics

Etymologically, genetics comes from the word 'genos' (Latin) means an ethnicity or origins.¹ Genetics is a study of inherited traits and their variation, how the nature of the offspring (heredity) was bequeathed to the children and grandchildren and

¹ Dr. Wildan Yatim., Genetika, (Bandung: Tarsito, 1996), p.1

also the variations that may arise in it. Genetics is different from genealogy. Genealogy is more emphasis on a relationship rather than the nature of inheritance². With the advent of tests that can predict genetic illness, people have even compared genetics to fortunetelling, but genetics is neither genealogy nor fortunetelling, genetics is the a life science.

Definitions of genetics according to several sources are:

- In the dictionary of biology, genetics is the study of genes and its influence³
- According to Ricki Lewis, genetics is the study of inherited traits and their variation.⁴
- Anna C. Pai said that genetics is a science of heredity; it's the study of inherited traits from one generation to the next generations.⁵
- According to Bambang Supriyadi, genetics is the branch of biology that studies the characters that decreased from parent to child⁶.
- According to Toegiono, genetics is the study of ways to pass on the nature of the parent to the offspring.⁷

² Ricki Lewis, *Human Genetics: Concepts and Application*, (New York: Mc. Graw Hill, 2005), p.2

³ M Ambercrombie, dkk, *Kamus Lengkap Biologi* (Jakarta: Penerbit Erlangga, 1993) p. 260

⁴ Ricki Lewis, *Human Genetics: Concepts and Application.....*, p.2

⁵ Anna C. Pai, *Dasar-Dasar Genetika, Ilmu untuk Masyarakat*, (1992, Jakarta: Penerbit Erlangga), p. 1

⁶ Bambang Supriadi, dkk, *Modul Biologi*, (Yogyakarta: Primagama, 1992), p.2

⁷ Toegiono, *Genetika I*, (Surakarta: Universitas Sebelas Maret, 1992), p.1

So, genetics is the study of heredity—the inborn factors, inherited from the biological parents, that affect development. When sperm and ovum unite, they endow the baby -to- be with the genetic makeup that influences a wide range of characteristics from color of eye and hair to health, intellect and personality.⁸

And because the characters that derived from parents contained in genes (and genes are also the cause of genetic variation), genetics can also be regarded as the science of genes.

2) GENES

Genes⁹ are units of heredity (descent). Genes control many traits, from hair color to the human character. There is a biochemical instruction that tells the cell as the basic unit of life how to manufacture certain proteins. These proteins ultimately underlie specific traits, they provide a great variety of characteristic that create much of our individuality, from our hair and eye color, to the shapes of our body parts, to our talents and personality traits. For example, proteins called keratins comprise our hair and fill our skin cells. One consequence of impaired keratins production is the scaly skin disease ichthyosis¹⁰

Chromosomes contains of DNA molecules¹¹

⁸ Diane E. Papalia, dkk, *Human Developmentt*, (New York: Mc. Graw Hill, 2004),..p. 65

⁹ Gene is small segment of DNA located in definite positions on particular chromosomes.

See Diane E. Papalia., dkk, *Human Development.....*, p. 65

¹⁰ Lewis, Ricki, *Human Genetics: Concepts and Application.....*, p.2

¹¹ DNA is the genetic material in all living cell. The structure of DNA resembles a long, spiral ladder made of four chemical units called *bases*. The bases –

(*deoxyribonucleic acid*) that looks like the yarn. Each chromosome contains thousands of genes which is located in specific places. In each gene, there are the combinations of four elements, namely DNA amino acids: adenine, thymine, cytosine, and guanine (ATCG) that form a chemical code that plays a role in determining the specific protein synthesis. Later, these proteins will affect the overall structure and biochemical characteristics of organism.



Inherited traits:
Gen control many familiar traits, from hair colour to athletic powers

The main purposes of Genes:

1. Regulating the growth and metabolism of individual
2. Submitting to the next generation of genetic information.¹²

In the era of Gregor Mendel, genes are often referred as the determinant factor or element or a determinant.

Thomas Hunt Morgan (1866-1945), a geneticist and embryologist of United States, expressed his opinion that the gene is the substance of heredity, it is a unity of chemistry that have characteristics as follows:

adenin, thymine, cytosine, and guanine- are known by their initials: A,T,G, and C. They pair up in four combinations: AT, TA, CG, and GC and coil around each other. The sequence of 3 billion base pairs constitutes the genetic code, which determines all inherited characteristics. see Papalia, Diane E, dkk, *Human Development*.....p. 65-66

¹² http://www.crayonpedia.org/mw/Gen_Sebagai_Substansi_Hereditas_12.1, 12/01/2011

- 1) Gene is a compact particle in the chromosome.
- 2) Gene contains genetic information.
- 3) Genes can duplicate itself in the phenemenons of mitosis and meiosis; means that genes can be split into two, one is as like as the other, therefor it can deliver genetic information to the next cell generation.
- 4) Each gene occupies a particular place in the chromosomes, which occupied a special location of genes on the chromosome called locus genes.¹³

Every cell in the normal human body except the sex cells has 23 pairs of chromosomes -46 in all. Through a type of cell, division called meiosis, each sex cells, or gamete (sperm or ovum), ends up with only 23 chromosomes- one from each pair. Thus, when sperm and ovum fuse at conception, they produce a zygote with 46 chromosomes, 23 from the father and 23 from mother.¹⁴

A pair of chromosomes is a fellow homologous, means the locus of homologous chromosomes also contained the corresponding genes. The corresponding genes in the corresponding loci on homologous chromosomes called alleles (gene pair). So the gene is the smallest unit of heredity nature (heredity substance) which is about 40-50 milimicron.

¹³ http://www.crayonpedia.org/mw/Gen_Sebagai_Substansi_Hereditas_12.1, 12/01/2011

¹⁴ Diane E. Papalia, dkk, *Human Development*.....p.66

Genes do not do their work automatically. They spring into action when conditions call for the information they can provide. Genetic action that triggers growth of body and brain is often regulated by hormonal levels, which are affected by such environmental conditions as nutrition and stress. Thus, from the start, heredity and environment are interrelated.¹⁵

3) Application of Genetics

Barely a day goes by without some mention of genetics in the news. Genetics is impacting many areas of our lives. Genetics greatly influence our lives from our health care choices, to what we eat and wear, to unraveling our pasts, and controlling our future. Some areas of the application of genetics:

a) Establishing Identity and Origin

Comparing DNA sequences to establish or rule out identity, relationship, or ancestry is becoming routine. This approach called DNA profiling¹⁶. It can be applied on some cases such as:

¹⁵ Diane E. Papalia., dkk, *Human Development*.....p.66.

¹⁶DNA profiling is The activity of collecting DNA to get information about someone, especially criminal, in order to give description of them. Its also helps adopted individuals locate blood relatives. The Kinsearch Registry maintainance a database of DNA information on people adopted in the United States from China, Rusia, Guatemala, and Shout Korea, the sources of most foreign adoption. Adopted individuals can provide a DNA sample and search the database by country of origin to find siblings.

DNA profiling can also trace origins for organism other than human. For example, the researchers analyzed DNA from the leaves of 300 varieties of winw grapes, in search of the two parental strain that gave rise to the sixteen major types of wine

- a. Forensic, such as in the investigation to know the evil by using fingerprint and DNA of evil doer.
- b. Rewriting history, usually it is used to make family trees. DNA can help to flash out details of history, and something spring surprises.
- c. Tracing Origins usually used to know the origin of an ethnic.

b) Health Care

Inherited illness caused by a variant in a single gene differs from other types of illness in several ways. And the differences between genetic diseases and another diseases are:

- One can predict recurrence risk in other family members
- Predictive testing is possible
- Different populations may have different characteristic frequencies.
- Correction of the underlying genetic abnormality may be possible.¹⁷

A few genetic diseases can be treated. Supplying a missing protein can prevent some symptoms, such as providing a clotting factor to a person who has the bleeding disorder hemophilia.¹⁸

grapes existing today. See, Lewis, Ricki, *Human Genetics: Concepts and Application*,....., p.11-13

¹⁷ Lewis, Ricki, *Human Genetics: Concepts and Application*....., p.14

¹⁸ One example of genetic application in health care is using gene therapy. Gene therapy is the insertion, alteration, or removal of genes within an individual's cells and biological tissues to treat disease. It is a technique for correcting defective genes that are responsible for disease development. The most common form of gene therapy involves

Balancing the perceived risk to privacy that genetic test present are the possibilities that such test can lower health care costs. If people know their inherited risk, they can take measures to forestall or ease symptoms that environmental factors might trigger- for example by eating healthy foods, not smoking, exercising regularly, avoiding risk behavior, having frequent medical exams, and beginning treatment earlier. Genetics test can also enable people to make more informed reproductive decision. People who know that they can transmit an inherited illness may elect not to have children, or to use one of the assisted reproductive technologies.¹⁹

c) Agriculture

Both traditional and biotechnical apply genetic principles. Traditional agricultures are the controlled breeding of plants to select individuals with certain combinations of useful inherited traits such as seedless fruits. Biotechnology is the use of organism to produce goods (including food and drugs) or services, and it's an ancient art as well as modern sciences.²⁰

So, by genetics, we can learn how to find the best seeds of a plant, such as a transgenic organism that contains several genes

the insertion of functional genes into an unspecified genomic location in order to replace a mutated gene, but other forms involve directly correcting the mutation or modifying normal gene that enables a viral infection. Although the technology is still in its infancy, it has been used with some success.

http://en.wikipedia.org/wiki/Gene_therapy on 5/12/2011, 9:12 AM

¹⁹ Lewis, Ricki, *Human Genetics: Concepts and Application.....*, p.14-15

²⁰ Lewis, Ricki, *Human Genetics: Concepts and Application.....*, p.15

from different species and it also can be used to get double production in the harvest.²¹

d) Animal Healthy and Husbandry

Beside in order to know the descent abnormalities and nature exploratory of the offspring to determine the origin of livestock, genetics can also be used to obtain good seeds, such as: to produce more meat, milk, eggs, and plants become more resistant to disease.²²

e) Ecology

In metagenomics²³, DNA that collected from some habitat used to reconstruct the ecosystem.

²¹ Dr. Wildan Yatim, *Genetika.....*, p.3

²² Dr. Wildan Yatim, *Genetika.....*, p.2

²³ Metagenomics is the study of metagenomes, genetic material recovered directly from environmental samples. Applications of Metagenomics:

Many microorganisms have the ability to degrade waste products, make new drugs for medicine, make environmentally friendly plastics, or even make some of the ingredients of food we eat. By isolating the DNA from these organisms, it provides us with the opportunity to optimize these processes and adapt them for use in our society. As a result of ineffective standard laboratory culture techniques, the potential wealth of biological resources in nature (like microbes) is relatively untapped, unknown, and uncharacterized.

Metagenomics represents a powerful tool to access the abounding biodiversity of native environmental samples. The valuable property of metagenomics is that it provides the capacity to effectively characterize the genetic diversity present in samples regardless of the availability of laboratory culturing techniques. Information from metagenomic libraries has the ability to enrich the knowledge and applications of many aspects of industry, therapeutics, and environmental sustainability. This information can then be applied to society in an effort to create a healthy human population that lives in balance with the environment. Metagenomics is a new and exciting field of molecular biology that is likely to grow into a standard technique for understanding biological diversity.

f) Genetics in Global Perspective

Human genome information²⁴ has tremendous potential but must be carefully managed. As like as genetic engineering²⁵ that will be very useful if used correctly and will certainly be very dangerous if the use is wrong.

B. PATTERN OF GENETIC TRANSMISSION/HEREDITY

a) Theory of The Origin of Human

Before the 17th century, the famous theory of the origin of life is the theory of "generatio spontania". Its theory said that life arose spontaneously. The famous proof of this theory is about the appearance of maggots on raw meet.²⁶

18th and 19th century, by the improvement of microscopes and scientific techniques, it is known that there were no spontaneous lives, which all living things come from its predecessor living things. But how the way of the parent to offspring inheritance is not known yet, there is no theory about the process of inheritance.²⁷

²⁴ Human genome information can ultimately benefit for everyone. Consider drug development. Today, there are fewer than 500 types of drugs. Genome information from human and our pathogens and parasites is revealing new drug targets. Global organizations, including the United Nation, can share new diagnostic test and therapeutics that arise from genome information. See Lewis, Ricki, *Human Genetics: Concepts and Application*....., p.16-17

²⁵ Genetic engineering is (the science of) changing the structure of the genes of a living thing in order to make it healthier or stronger or more useful to humans

²⁶ Anna C. Pai, *Dasar-Dasar Genetika: Ilmu Untuk Masyarakat* (Jakarta: Erlangga, 1998),p.3

²⁷ Anna C. Pai, *Dasar-Dasar Genetika: Ilmu Untuk Masyarakat*p. 2-3

The old concept of Heredity

Greek philosophers have many different ideas about heredity. Theophrastus proposed that male flowers caused the female flower to ripen, Hippocrates suspected that the "seed" is produced by various part of the body and inherited at the time of conception, Aristotle said that males and females cement mixed at fertilization, while Aeschylus, at 458 BC put the idea forward that the males are the real parents and female is only the "nurse for the young life sown within her"²⁸

Various mechanisms of heredity proposed without tested or quantified expediently. These mechanisms include a blending inheritance, and inheritance of acquired characteristics. However, people were able to develop domestic breeds of animals and plants artificial selection. The inheritance of acquired traits also formed the part of early Lamarckian ideas on evolution.

In the eighteenth century, a microscopist, Antoine van Leeuwenhoek (1632-1723) found "animalcules" in the sperm of human and the other animals. This discovery became the basis of the theory of "spermists" which stated that there is "little man" (homunculus) inside a sperm and the only contribution made by the woman is the womb in which homunculus grew and prenatal influences of the womb. Other theories be in contradictories with that theory, its "Ovists" which suspected that women who keep a small man in the ovum. Ovists thought women carried eggs

²⁸ <http://en.wikipedia.org/wiki/Heredity> on 12/05/2011

containing boy and girl children, and that the gender of the offspring was determined well before conception.²⁹

Pangenesis is an idea which stated that men and women formed a "pangen" in each organ of man. This concept appeared in ancient Greece and influenced the life sciences about a hundred years ago. The term "blood ties", "pure blood", and "royal blood" are the remnants of Pangenesis theory. In 1870 Francis Galton, a cousin of Charles Darwin did experiments that deny Pangenesis.³⁰

b) Human Development

Human reproductive system consists of the testes for males and ovaries for women.³¹ The formation of new individual begin from the meeting of sperm and ovum in the uterus. Fertilization³² will occur in the fallopian tube and there would be only one sperm from the thousands of sperm that successfully fertilize the egg. Only sperm that have undergone capacitating that can cross the pellucid zone and into vitellus ova. After that, the pellucid zone changes, so that ovum cannot be passed by other sperm.

Fertilization is most likely if intercourse occurs on the day of ovulation or during the five days before. If fertilization

²⁹ <http://en.wikipedia.org/wiki/Heredity> on 12/05/2011

³⁰ Anna C. Pai, *Dasar-Dasar Genetika.....*,p.3

³¹ Lewis, Ricki, *Human Genetics: Concepts and Application.....*, p.48.

³² Fertilisation or conception is the process by which sperm and ovum combine to create a single cell called a zygote, which then duplicates itself again and again by cell division to become a baby. Diane E. Papalia, dkk, *Human Development*, (New York: Mc. Graw Hill, 2005), p.63

does not occur, the ovum and any sperm cells in the woman's body die. The sperm are absorbed by the woman's white blood cells and the ovum passes through the uterus and exists through the vagina.³³



Human Sperm:

- a. The structure of sperm that brings trait for the next generation
- b. Scanning electron micrograph of human sperm cells
- c. Homunculus, the illustration of sperm on 16th centuries.

When the sperm of men mixed with ovum (female egg cell), the essence of the baby to be born is formed. Single cell - known as a zygote³⁴ in the biological sciences- will directly begin to reproduce itself from one cell divides into two cells, two cells become four cells, four cells become eight cells, and so on until eventually it became a piece of meat. Guided by a blue print that is contained in the genes inherited from parents, and then these cells differentiate in accordance with the functions and aims of cell established.³⁵

c) Mendelian Inheritance

The first scientist who proved that the transfer of characters did not always doubt but it has a pola and can be

³³ Diane E. Papalia, dkk, *Human Development*....., p. 64

³⁴ One-celled organism resulting from fertilisation, see Diane E. Papalia., dkk, *Human Development*....., p.63

³⁵ Roni, Dr. Noor Rahman, dkk, *Rahasia dan Hikmah Pewarisan Sifat (Ilmu Genetika dalam Al Qur'an)*, 2010, Bandung: IPB Press, p.12-13

predicted is Gregor Mendel³⁶, a monk from Austria. He was the first man who triggers the law of classical genetics (19th century).³⁷

In his experiments Mendel used pea beans (ercis). Mendel succeeded to observe a range of characters of offspring from generation to generation and also made the calculation that he called as a determinant. Therefor Mendel called as 'the father of genetics' whom provided the basis of modern genetics.³⁸

Mendel discovered that when crossing white flower and purple flower plants, the result is not a blend. Rather than being a mix of the two, the offspring was purple flowered. He then conceived the idea of heredity units, which he called "factors",

³⁶ Mendel spent his early childhood in a small village in what is now the Czech Republic, near the Polish border. His father was a farmer and his mother was the daughter of gardener, so Mendel learned early how trend fruits trees. At age 10 he left home to attend a special school for bright students, supporting himself by tutoring. After a few years at a preparatory school, Mendel became a priest at the Augustinian monastery of St. Thomas in Brno. At this atypical monastery, the priests also the teachers, and they did the research in natural sciences. From them Mendel learned how to artificially pollinate crop plants to control their breeding.

At age 29, such an effective substitute teacher that he was sent to earn a collage degree. At the university of Vienna courses in the sciences and statistic fuelled his enduring interest in plant breeding and got him thinking about the experiment to address a question that had confounded other plant breeders –why did the certain traits disappear in one generation yet reappear in the next?– And to solve this puzzle Mendel bred hybrids and applied the statistics he had learned in collage.

From 1857-1863, Mendel crossed and cataloged some 24.034 plants, through several generation. He deduced that consistent ratios of traits in the offspring indicated that the plants transmitted distinct units or element. He derived two hypotheses to explain how inherited traits are transmitted. Mendel described his work to Brno Medical Society in 1865 and published it in the organization's journal the next year. See Lewis Ricki, *Human Genetics: Concepts and Application.....*, p.76

³⁷ Anna C. Pai, *Dasar-Dasar Genetika: Ilmu Untuk Masyarakat.....*p. 4

³⁸ Dr. Wildan Yatim, *Genetika.....*, p.3

one of which is a recessive characteristic and the other dominant. Mendel said that factors, later called genes, normally occur in pairs in ordinary body cells, yet segregate during the formation of sex cells. Each member of the pair becomes part of the separate sex cell. The dominant gene, such as the purple flower in Mendel's plants, will hide the recessive gene, the white flower. After Mendel self-fertilized the F1 generation and obtained the 3:1 ratio, he correctly theorized that genes can be paired in three different ways for each trait: AA, aa, and Aa. The capital "A" represents the dominant factor and lowercase "a" represents the recessive. (The last combination listed above, Aa, will occur roughly twice as often as each of the other two, as it can be made in two different ways, Aa or aA.)

Mendel stated that each individual has two factors for each trait, one from each parent. The two factors may or may not contain the same information. If the two factors are identical, the individual is called homozygous for the trait. If the two factors have different information, the individual is called heterozygous. The alternative forms of a factor are called alleles. The genotype of an individual is made up of the many alleles it possesses. An individual's physical appearance, or phenotype, is determined by its alleles as well as by its environment. An individual possesses two alleles for each trait; one allele is given by the female parent and the other by the male parent. They are passed on when an individual matures and produces gametes: egg and sperm. When gametes form, the paired alleles separate randomly so that each

gamete receives a copy of one of the two alleles. The presence of an allele doesn't promise that the trait will be expressed in the individual that possesses it. In heterozygous individuals the only allele that is expressed is the dominant. The recessive allele is present but its expression is hidden.

In brief, the theory of Mendel concluded that mother nature does not blend in the offspring. The resulting offspring have one trait parent (mother or father) and highly dependent on the most dominant genes between both mother or father. In early 1900, W.L. Johansson proposed the term gene to express the units of inheritance. Although Mendel did not call it genes, but the unit, but from Mendel we know the nature of dominant and recessive.³⁹

After did experiment, Mendel finally formulating important laws related to the crossbred between the different varieties with one trait. Explicitly the laws are:

- 1) Marriages between plants or animals from two different varieties will produce offspring that have similarity to their parent.
- 2) All individuals who are first descendants are always same.
- 3) If the resulting offspring have the same nature with one parent, so there is the dominance of the gene from one parent (the dominant law).
4. If there is dominance, then the resulting offspring have characteristics of 75% of the dominant parent gene, and the other

³⁹ Anna C. Pai, *Dasar-Dasar Genetika: Ilmu Untuk Masyarakat.....*p. 7

25% of the parent gene that has not dominant gene (law of segregation⁴⁰).

5) The combination that appears in the offspring can be various (Law of free assortment⁴¹).⁴²

The laws that has been found by Mandel is a basic of modern genetic, so the biologist agreed to give appellation `The Father of Modern Genetics` to him. And then after the findings of Mendel, genetics developed rapidly.⁴³

⁴⁰The Law of Segregation states that when any individual produces gametes, the copies of a gene separate so that each gamete receives only one copy. A gamete will receive one allele or the other. The direct proof of this was later found following the observation of meiosis by two independent scientists, the German botanist, Oscar Her twig in 1876, and the Belgian zoologist, Edouard Van Beneden in 1883. In meiosis, the paternal and maternal chromosomes get separated and the alleles with the traits of a character are segregated into two different gametes. <http://en.wikipedia.org/wiki/Genetics> on 23/05/2011

⁴¹ The Law of Independent Assortment, also known as "Inheritance Law" states that alleles of different genes assort independently of one another during gamete formation. While Mendel's experiments with mixing one trait always resulted in a 3:1 ratio between dominant and recessive phenotypes, his experiments with mixing two traits (dihybrid cross) showed 9:3:3:1 ratios. But the 9:3:3:1 table shows that each of the two genes are independently inherited with a 3:1 phenotypic ratio. Mendel concluded that different traits are inherited independently of each other, so that there is no relation, for example, between a cat's color and tail length. This is actually only true for genes that are not linked to each other. <http://en.wikipedia.org/wiki/Genetics> on 23/05/2011

⁴² Anna C. Pai, *Dasar-Dasar Genetika: Ilmu Untuk Masyarakat.....*, p. 4-13

⁴³ The development of genetics is often being a classic example of the using of scientific method in science. Here are the stages of the development of genetics:
 1859 Charles Darwin published The Origin of Species, as the basis of genetic variation.;
 1865 Gregor Mendel submit manuscripts Crossbreeding Experiments on Plants;
 1878 E. Strassburger provide an explanation of double fertilization;
 1900 The discovery of Mendel's work again separately by Hugo de Vries (Belgium), Carl Correns (Germany), and Erich vonT schermak (Austro-Hungary) ==> early classical genetics;
 1903 Chromosomes are known as a units of genetic inheritance;
 1905 British biologist William Bateson acquaintance the term 'genetics';
 1908 and 1909 the basis of population genetics theory by Weinberg (German physician) and separately by James W. Hardy (British mathematician) ==> early population genetics;
 1910 Thomas Hunt Morgan showed that genes located on chromosome, by using the fruit

-
- fly (*Drosophila melanogaster*) ==> the beginning of cytogenetic;
- 1913 Alfred Sturtevant create the first genetic map of a chromosome;
- 1918 Ronald Fisher (biostatistician from the UK) published *On the Correlation Between relatives on the supposition of Mendelian inheritance*, which ended the feud between the theories of biometry (Pearson et al.) And Mendel's theory as well as initiate synthesis of both ==> early quantitative genetics;
- 1927 Physical changes in genes called mutations;
- 1928 Frederick Griffith discovered a carrier molecule that can be moved inter bacteriae (conjugation);
- 1931 Crossing causing recombination;
- 1941 Edward Lawrie Tatum and George Wells Beadle showed that genes encoding proteins, ==> beginning of principal theory of genetics;
- 1944 Oswald Theodore Avery, Colin MacLeod and Maclyn McCarty isolated DNA as the genetic material (they called transformation principle);
- 1950 Erwin Chargaff showed the general rules that apply to the four nucleotides in nucleic acids, for example, adenine tend to be as much as thymine;
- 1950 Barbara McClintock discovered transposons in maize, tend to be as much as thymine;
- 1950 Barbara McClintock discovered transposons in maize;
- 1952 Hershey and Chase proved that the bacteriophage genetic information (and all other organisms) is a DNA;
- 1953 The puzzle of DNA structure answered by James D. Watson and Francis Crick, it's the double helix, based on ray diffraction images of Rosalind Franklin's on X DNA ==> beginning of molecular genetics;
- 1956 Jo Hin Tjio and Lev Albert ensuring that the human chromosome amount to 46;
- 1958 Meselson-Stahl experiment showed that the DNA is duplicated (replicated) in semiconservatively;
- 1961 genetic code arranged triplet;
- 1964 Howard Temin showed using the virus of RNA that the basic theory is not always apply;
- 1970 Restriction enzymes found in *Haemophilus influenzae* bacteria, its possible to do cutting and joining of DNA (see also RFLP) ==> the beginning of modern biotechnology;
- 1977 DNA is sequenced by Fred Sanger, Walter Gilbert and Allan Maxam. Tim Sanger successfully sequenced the entire genome Bacteriophage Φ -X 174, a virus ==> beginning of genomics;
- 1983 Propagation (amplification) of DNA can be done easily after Kary Banks Mullis discovered polymerase chain reaction (PCR);
- 1985 Alec Jeffreys invented genetic fingerprinting.
- 1989 was the first sequenced of the human gene, encoding the protein of CFTR, cause cystic fibrosis;
- 1989 Laying a strong foundation for statistical analysis of quantitative trait loci (QTL analysis);
- 1995 *Haemophilus influenzae* genome sequencing, which became the first sequenced genome of a free living organism;
- 1996 The first sequencing of eukaryotes: *Saccharomyces cerevisiae* yeast;
- 1998 The first sequencing of multicellular eukaryotes, the nematode *Caenorhabditis elegans*, was announced;

a. Dominant dan Resessif Inheritance

Can you curl your tongue? If so, you inherited this ability through dominant inheritance. If your parents can curl their tongues but you cannot, recessive inheritance occurred. How do these two types of inheritance work?⁴⁴

Mendel stated that each individual has two factors for each trait, one from each parent. The two factors may or may not contain the same information. If the two factors are identical, the individual is called homozygous for the trait. If the two factors have different information, the individual is called heterozygous. The alternative forms of a factor are called alleles. The genotype of an individual is made up of the many alleles it possesses. An individual's physical appearance, or phenotype, is determined by its alleles as well as by its environment. An individual possesses two alleles for each trait; one allele is given by the female parent and the other by the male parent. They are passed on when an individual matures and produces gametes: egg and sperm. When gametes form, the paired alleles separate randomly so that each gamete receives a copy of one of the two alleles. The presence of an allele doesn't promise that the trait will be expressed in the individual that possesses it. In heterozygous individuals the only

2001 early draft of human genome sequence was released with the start of the Human Genome Project;

2003 Human Genome Project (Human Genome Project) completed 99% work on the date (14 April) with an accuracy of 99.99%.

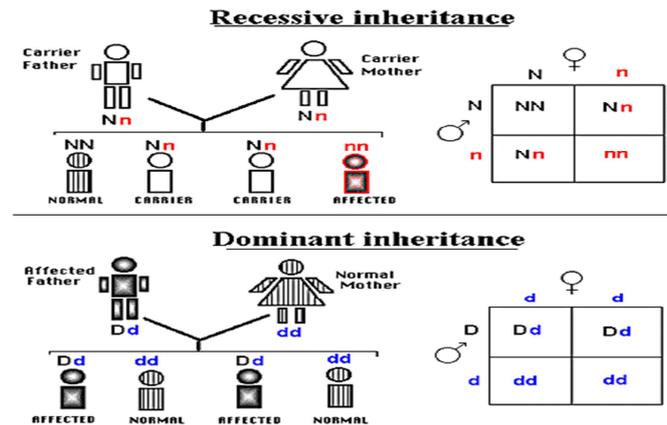
Lamarck, J-B (2008). In Encyclopædia Britannica. Taken from Encyclopædia Britannica Online on 16 March 2008. [Http://www.wikipedia.wiki.genetika](http://www.wikipedia.wiki.genetika). On 01/01/2011

⁴⁴ Papalia, Diane E., dkk, *Human Development*.....p.68

allele that is expressed is the dominant. The recessive allele is present but its expression is hidden.⁴⁵

So, in a crossing, the result of offspring will have some nature that appear or does not appear (hidden) from one of the traits of its parent. Nature that appears in the offspring called the dominant trait. On the contrary nature that does not show up or hidden because of being defeated by the nature of her partner called the recessive trait.

The differences between these two inherited traits are:



1. dominant inherited:

- a. the mixing between the two gametes which both contain the abnormal gene will result homozygous abnormal individuals (diseased).

⁴⁵ http://en.wikipedia.org/wiki/Mendelian_inheritance on 12/05/2011

- b. the mixing among the gametes that contain the abnormal gene with a gamete containing the normal gene would result heterozygous abnormal individuals (diseased).
 - c. the mixing between the two gametes which both contain a normal gene would result in individuals homozygous normal.
2. resesif inherited:
- a. the mixing between the two gametes which both contain a normal gene would result in homozygous normal individuals.
 - b. the mixing between the gametes containing the normal gene with gametes that contain the abnormal gene will produce a normal heterozygous individuals, often called the carrier, which is an individual that is normal phenotypes, but actually contain the gene for a disease / abnormal gene.
 - c. Unity between the two gametes that both contain the abnormal gene will result in individuals homozygous abnormal (diseased)

Recessive defects are expressed only if a child receives the same recessive gene from each biological parent. Defect transmitted by recessive inheritance are more likely to be lethal at an early age than those transmitted by dominant inheritance. If dominantly transmitted defect killed before the age of reproduction, it could not be passed on the next generation and therefore would soon disappear. A recessive defect can be transmitted by carriers who do not have the disorder and thus may

live to produce.⁴⁶

Some traits are not transmitted by simple dominant or recessive inheritance. *Codominance* occurs when neither of two alleles is dominant, and the resulting trait reflects the influence of both. For example, the blood type AB is a combination of allele for types A and B.⁴⁷

b. Inbreeding

The meaning of inbreeding is prohibited sexual relations because of the existence of family relationships⁴⁸. This sexual contact can occur between father and daughter, mother and son, between man and his sister, a certain cousin, stepfather and daughter, and many more are prohibited in religion and culture. Incestuous sexual contact can be prohibited in culture, can also prohibited healthily.

Prohibition by the culture is usually caused by several things that happen depending on each culture, which is sometimes difficult to understand why this could occur. However, inbreeding can happen because of disorder in the relationship between one with another in a family. For example, brothers who had long been separated, which subsequently met and married but do not know if they really are brothers.⁴⁹

⁴⁶ Diane E. Papalia, dkk, *Human Development*.....p. 71

⁴⁷ Diane E. Papalia, dkk, *Human Development*.....p.68

⁴⁸ http://en.wikipedia.org/wiki/Cousin_marriage on 02/012011

⁴⁹ <http://bidansmart.wordpress.com> on 8/2/2010

Long time ago, among the kings of Egypt, Ireland, and Inca in South America, marriage between siblings become common thing. Even this is considered to purify the blue blood. In the Pharaoh Dynasty in Egypt, Aahmes married with Aahmes-Neftari, his sibling. Their children, Amenhotep I and Aahotep also married with their siblings. In the tradition of the Jews, inbreeding also becomes common. But after Moses become prophet, inbreeding was prohibited.⁵⁰

According to B. Malinowski, an anthropologist, in some tribes on Malaysian and Indonesian islands we will still find the tradition of inbreeding. In Africa, the South Pacific island, there is many mom married with her son or father with his daughter until nowadays.⁵¹

According J.A. Fraser Roberts and Marcus E. P, in the inheritance of dominant diseases, in inbreeding -marriage between two individuals who still have consanguinity- will only reduce the number of individuals who are sick. It will be different from the inheritance of recessive diseases, because in this inheritance, the marriage only will produces more the sick man than the marriage between two heterozygous (carrier) individuals.⁵²

But the disease that is a dominant inheritance is very rare in daily life, which is more common, is the transmission of diseases that are recessive inheritance. In a recessive inheritance,

⁵⁰ Dr. Wildan Yatim, *Genetika*, (Bandung: Tarsito, 1996), p.316

⁵¹ Dr. Wildan Yatim, *Genetika.....*, p.316

⁵² Roberts, Fraser J.A & Marcuse E. P, *Genetika Kedokteran.*, (Jakarta: EGC, 1995) p.51

if there is marital relations/marriage between individual carriers who still have blood relationship, then it is probable that he married individuals who are also carriers. This is related to inheritance from both parents, and blood relationship is more likely to give the same gene each other compared with those who have no blood relationship.

Based on research results, the levels of inbreeding can be calculated by calculating the coefficient value of inbreeding on an individual by using his family genealogy. For example, the coefficient between siblings amounted to $\frac{1}{2}$ (50%). While inter-cousin inbreeding coefficient amounted to $\frac{1}{4}$ (25%). This number means that the opportunity to obtain the same gene among siblings was 50%. The similarity of our genes with the biological mother and father is 50%, because half of our genes come from our father and 50% more than mothers. In the early stages of inbreeding, usually it will result the better performance of offspring because of the good qualities of elders incorporated and refined on the individual. In the next stages, the inbreeding will reduce the performance such as reproduction stage decreasing, mortality increasing, production decreasing, and increasing of occurrence of defects and abnormalities.⁵³

⁵³ Roni, Dr. Noor Rahman, dkk, *Rahasia dan Hikmah Pewarisan Sifat (Ilmu Genetika dalam Al Qur'an)*, 2010, Bandung: IPB Press, p.84

d) Sex Determination

Sex determination is the mechanism by which an organism develops as a male or a female. In many species, males and females differ in the types of sex chromosomes that they have. These chromosomes may determine sex because they include genes that direct development of reproductive structures of one sex, while suppressing development of structures of the other sex.⁵⁴

In many villages in Nepal, it's common for a man whose wife has borne no male babies to take a second wife. In some societies, a woman's failure to produce sons is justification for divorce. The irony in the beliefs about conception underlying these customs in male-dominated societies is that it's the father's sperm that determines a child's sex.⁵⁵

So in ancient times, many nations had a presumption that the mother determines the sex of offspring. Many wives are often blamed for not being able to give only male offspring to their husbands. With the development of genetics, we can now know how the transmission of sex determination works in humans.⁵⁶

At the moment of conception, the 23 chromosomes from the sperm and 23 chromosomes from the mother's ovum form 23 pairs. Twenty-two pairs are autosomes, chromosomes that are not related to sexual expression. The twenty-third pair are sex chromosomes-

⁵⁴ Lewis, Ricki, dkk, *Life*, (New York: Mc. Graw Hill, 2007), p.212

⁵⁵ Diane E. Papalia, dkk, *Human Development.....*, p. 66

⁵⁶ Anna C. Pai, *Dasar-Dasar Genetika: Ilmu Untuk Masyarakat....*, p.54-55

one from the father and one from the mother- that give the baby's sex.⁵⁷

Chromosome is a major element in the determination of gender. Two of the 46 chromosomes that determine the structure of a human being are expressed as sex chromosomes. Both are called the XY chromosomes in males and XX in females. This is because the shapes of the chromosomes are similar to these letters. The Y chromosome carries a gene that indicates the type of men, whereas the X chromosome carries a gene that indicates women.⁵⁸

One active field of medical research related to the effort to distinguish sperm that contain X from Y chromosomes. There were reports about the effects of uterine environment which alkalis or acids that support one or the other, and the differential displacement of the sperm that contain X or Y in an electric field.⁵⁹

C. GENETICS AND BEHAVIOUR

1. Genes and Environment

In our daily life, we often meet a person whose body is tall, medium, or even dwarf. We also often meet with people who are thin or fat, people who have straight or curly hair, healthy people or people with diseases or disorders such as

⁵⁷ Diane E. Papalia, dkk, *Human Development*.....,p.66

⁵⁸ Anna C. Pai, *Dasar-Dasar Genetika*..., p. 54

⁵⁹ Anna C. Pai, *Dasar-Dasar Genetika*..., p.56

albino, color blindness, and others. If we look carefully, we can observe some the following story:

- (1). Case I: the child is high, and it's usually born from the couples who either parents or one parent is high, it's also happen on fat or dwarf child. Children who has color blindness, usually in his family's genealogy is also has colour blindness.
- (2). Case II: children whose curly hair or straight hair, usually have a mother or father who is also curly or straight hair, except the person who originally straight then he go to the salon and then he changed her hair into curls, or people who had curly then her hair straighted to become straight.
- (3). Case III: people who always do the muscles sport will have strong and sturdy body, for examples bodybuilders, although his parents did not have strong muscles.

In the science of genetics known the term of phenotypes, that is the nature of offspring that can be seen or observed, such as skin color, face shape, body size, etc⁶⁰. Beside, genotypes is the genetic composition of which was conceived by an individual⁶¹ or nature that are not visible and its permanent on a single individual⁶². from these 3 cases above, we may conclude that:

Case I, which appeared absolute phenotypes that determined by genetic factors inherited from her parents, if either parents or one

⁶⁰ Toegiono, *Genetika I*, (Surakarta: Universitas Sebelas Maret, 1992), p.7

⁶¹ Toegiono, *Genetika I.....*, p.3

⁶² Supriadi, Bambang, dkk, *Modul Biologi*, (Yogyakarta: Primagama, 1992),

of them, or in the genealogy of his family did not carry the gene, the traits may not arise to her son.

Case II can be concluded that the phenotypes determined not only by parents genes but also by the environment. This means that even if a child does not carry the gene curly hair from her parents but she could turn into curly hair straight using a particular drug in the salon.

Meanwhile in the case III: the phenotypes seen absolutely determined by the environment. Another example is people are expert in certain skills that he acquired from the training since childhood, although these skills are not owned by her parents.

According J.A. Fraser Roberts and Marcus E. P., a trends or characters can be classified as follows⁶³:

1. In people who only have a certain genetic constitution, and this characters are always happening and only happens to those people. So this character is determined by genetic factors completely.
2. In people who have certain genetic constitution, but the character was not found on all people with the genetic constitution, so the cooperation of environmental effects required for the appearance of the character.
3. in people with the genetic constitution and with the same frequency on constitutisgenetic. So that character is determined by the environment perfectly.

⁶³ Roberts, Fraser J.A & Marcuse E. P, *Genetika Kedokteran. Alih bahasa Hartono*, (Jakarta: EGC, 1995) p.7

2. Behavioural Genetics

Behavioural genetics is the field of study that examines the role of genetics in animal (including human) behaviour. Often associated with the "nature versus nurture" debate, behavioural genetics is highly interdisciplinary, involving contributions from biology, genetics, ethology, psychology, and statistics. Behavioural geneticists study the inheritance of behavioural traits. In humans this often use the twin study or adoption study.⁶⁴

In genetics, the expression of a trait is a combination of genes and environmental factors. Or, phenotype is the result of genotype and environment⁶⁵. Although the gene is the blue print, but its expression is largely determined by environmental factors. Environment can trigger gene expression and also suppress gene expression. So, even though a person is born with has a gene that will be made him as a criminal because of the aggressiveness genes, but if the environment do not support, or he get good

⁶⁴ [http://www.wikipedia.wiki.behavioural genetics](http://www.wikipedia.wiki.behavioural%20genetics). On 24/10/2011

⁶⁵ For example: lets say that Steven has inherited musical taleent. If he takes musical lesson and practice regularly, he may delight his family with his performance. If his family likes and encourages classical music, he may play Bach preludes, if the other children on his block influence him to prefer popular music, he may eventually form a rock group. However, if from erly chilhood he is not encourage and not motivated to play music, and he have no access to a musical instrument or to music lessons, his genotype for musical ability may not be expressed(or may be expressed to a lesser extent) in his phenotype. Some physical characteristics (including height and weight) and most psychological characteristics (such as intelligence and personality traits, as well as musical ability) are products of multifactorial transmission. See Diane E. Papalia, dkk, *Human Development.....*,p.69

educations, then the gene expression will be depressed, then he would be a good person.⁶⁶

In the development of genetics, at 1980-1990 era, has found what we called as plasticity genes that have responsibility for the phenotype changing if an individual exposure in different environments.⁶⁷

Some characteristics influenced by heredity and environment:

- Physical and Physiological Traits
- Intelligence and School Achievement
- Personality
- Psychopathology⁶⁸

So can we conclude that human character can be influenced by several factors⁶⁹:

⁶⁶ Roni, Dr. Noor Rahman, dkk, *Rahasia dan Hikmah Pewarisan Sifat (Ilmu Genetika dalam Al Qur'an)*, 2010, Bandung: IPB Press, p.18

⁶⁷ Roni, Dr. Noor Rahman, dkk, *Rahasia dan Hikmah Pewarisan Sifat.....*, p.19

⁶⁸ See Diane E. Papalia, dkk, *Human Development.....*, p.82-86

⁶⁹ In the psychological perspective, there are two conflicting trends in determining which factors are more dominant between in shaping the human personality.

1). The trend of nativism that pioneered by Schopenhauer (1788-1860). It argued that internal factors are stronger or more dominant than the external factors. This trend is supported by naturalism pioneered by JJ Rousseau. The assumption of this trend is that the child and parents have many similarities both physically and psychologically. This trend also called as pessimistic and deterministic

In daily life we often find the people who live with the potential of nature, a talent that has carried since birth, which is difficult once modified by any influence. As an example is the prophet Muhammad, since he was young until being a messenger of God, he always avoided from the influence of the jaahiliyyah.

2). On the other hand, the trend of empiricism that pioneered by John Locke (1632-1704) with the theory of Tabula Rasa argues that the soul of a child from birth is still clean as an empty box. And will contain if it receives something from the outside.

1. Internal factors, are the factors that have been brought by human since in the mother's womb. It's in the form of seedlings, seeds, genes, and also the basic-skills which in Islam is called the potential disposition (*fitrah* potention).
2. External factors are environmental factors.

Both these factors give a huge influence for the development and formation of human characteristics and personality.

It's because influence from outside is stronger than human nature. It psychological assumption is every humans are born without anything, as like as a white paper (*tabula rasa*) that can be inscribed with whatever He wants. Embodiment of his behavior is determined by the environment. Baby will have the same tendency, feeding if his lips in contact with the breast of his mother, crying when thirsty and sick, and breasting when hungry. This trend is known as an optimistic and positivist trend.

In response to these two trends, W. Stern (1871-1929) proposed his famous theory; it's the theory of convergence. According to this theory, internal and external factors actually fused into one. Convergence is the interaction between heredity factors and environmental factors in the development of behavior. Heredity would not develop properly if not given the stimulation of environmental factors. In the contrary, environmental stimuli will not develop without based on heredity factor. Hence the determination of one's personality is determined by the integral work between internal factors (inherited potential) and external factors (environmental education). See Al Banjari, Rahmat Rmadhana, *Membaca Kepribadian Muslim Seperti Membaca Al Qur'an*, (Yogyakarta: DIVA Press, 2008), p. 27-32