RECESSIVE MUTATIONS AND SPERM BANKS OF GENIUS-POINTS OF VIEW

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"Gray matter"is one of the

most expensive goods in the world, which has long been bought and valued. In general, the intelligence quotient (IQ) is 3% of people over 130, and of these in a minority of 0.1% is 180, this category includes geniuses or gifted.

In 1776, Lazzaro Spallangoni discovered that human sperm could be frozen and "revived", and in the nineteenth century, biologists and geneticists, including H.J. Muller developed the storage of sperm fluid in a "sperm bank", this innovation being followed by the invention of intrauterine insemination techniques and, later, that of "in vitro" fertilization.

Initially, these procedures were used in patients of both sexes with genital diseases, such as oligospermia (for example, in men with vasectomies), with surgical pathology of the genitals or in infertility secondary to endometriosis, thus developing a prolific period of human reproduction.

In addition, it was requested to store and preserve the sperm of Nobel laureates (such as Paul Smith), in the hope of obtaining genius family descendants or for the optimal phenotypic qualities transmitted to the child, respectively a certain height, harmonious and/or athletic physique, blue eyes, blond appearance and so on.

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The sperm donor must meet several conditions, among which the age under 40 is required, knowing that, above a certain limit, advanced age is a mutagenic factor, especially for chromosomal abnormalities.

It is noteworthy that in humans there is no "parthenogenesis" (the formation of an embryo and the development of an organism in the unfertilized female gamete, as it exists in some lower species), the product of conception being the result of the fusion of the two gametes, respectively the fertilization of the female gamete. the ovum) by the male (sperm), in which the resulting genetic material gives rise to the product of conception.

The question is: if, in the family of a brilliant man, the resulting child has a normal or lower IQ, can his "gifted" sperm give another woman a prodigy child?

It was not possible to achieve geniuses "in series", with a single mother or with several mothers. The brilliant individuals who were born so far were randomly "produced". However, some situations of personalities with valuable descendants were also described and some investigations showed that talent is inherited, with families of gifted people having as examples: Bernoulli-mathematician, Strauss or Bach.

Some brilliant personalities manifest themselves early (Gauss-mathematician at the age of 3, Ampère- at the age of 4, Mozart- at 5 years, Haydn- at 6 years), instead others became active and, therefore, famous, after the age of 50- 60-70 years.

The genetic universe of the individual is a mosaic of dominant and recessive genes; the gene that manifests itself in heterozygous form is of dominant type, and the one that expresses itself in homozygous form is of recessive type. Very rarely, for unknown reasons, the carrier of a genetic mutation can be phenotypically healthy, but to procreate abnormal children.

According to several authors, such as Lombroso, geniuses also associated some pathological features, such as epilepsy-Caesar, Flaubert, Molière, Paganini, others were stuttering-Darwin or Aristotle, or, according to Ernst Kretschmer, had borderline mental illness.

Hereditary traits are controlled by genes made up of DNA (Johanssen W. 1909) located in chromosomes. The gene is the fundamental element of the hereditary chromosome. A chromosome consists of two molecules of DNA (chains of nucleotides, rolled around each other) and proteins, and DNA is the support of heredity. The DNA molecule comprises segments that correspond to a certain hereditary character.

According to J. Watson, the two strands of the DNA molecule can be rolled to the right-helix "right" or to the left, helix "Z" (DNA-Z),(Fig.1).

According to the theory of heredity, a genetic character can be **dominant** (it manifests in the offspring, even if it is transmitted only **by one parent**), respectively it can be **recessive,** if it is transmitted by both parents, so carried by both homologous genes.

Mutations can occur due to the action on the genome of some physical, chemical or biological factors.

Mutations transmitted from parents to children are called hereditary mutations or germline mutations, because they are present in germinal cells.

In humans, germline mutations are double in men compared to women,due to numerous divisions during spermatogenesis. This type of mutation cannot be treated. It will be present in every cell in the body of the affected person.

Mutations that occur only after fertilization are called new mutations. New mutations can explain genetic disorders if a child has a mutation in each cell, but has no other such cases in the family.

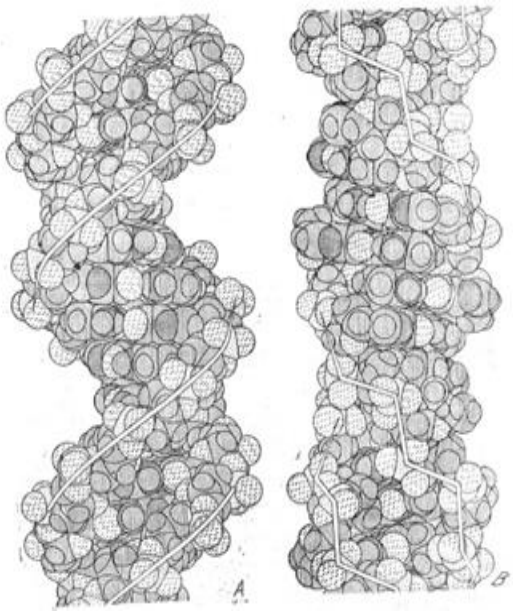

Fig.1: The DNA molecule; Watson-Crick model (A) and Z-DNA (B) - after Maximilian C.

From the point of view of their significance in the organism's relations with the environment, as well as in the process of evolution (so as an individual phenomenon), the variations can be divided into two main categories:

• gametic mutations (germinal) - changes in DNA in cells destined to become gametes or zygotes, which subsequently spread to the whole individual, and, therefore, and only in this way, can be passed on to offspring. Any mutation in single-celled organisms has the role of gametic mutation.

• somatic mutations - changes in DNA in somatic cells, which form the body's tissues. Variations in somatic tissues do not spread to many other cells, and do not reach other tissues (exception - cancer). These mutations exist until the cell dies and are not passed on to offspring.

Gametic mutations are the engine of evolution, while somatic mutations are irrelevant in this area, but their understanding is very important for studying the aging process and cancer.

GENIC mutations are the only category of mutations that affect the structure of DNA, namely, they usually cause changes in the structure of the genetic code.

CHROMOSOMIAL mutations do not usually affect the structure of genes but especially the connections between them. There are restructurings of chromosomes as a result of which whole groups, larger or smaller, of genes are moved from one chromosome to another.

Genetic mutation is "a detectable and hereditary change in genetic material" (Hugo de Vries), followed by detrimental effects. Unpredictable accident, genetic mutations are random. There are gene mutations - the most common, as well as chromosomal and genomic mutations.

If the structure of introns (non-informational nucleotide sequence of a gene) changes

frequently without reshaping with obvious consequences, exon mutations (informational sequence of a gene encoding a certain part of the final synthesized ribonucleic acid) are subject to natural selection and are clinically detectable. The mutation, through its direct consequences, affects the person through the presence of accumulated mutations in the genetic reservoir (Maximilian C.), sometimes exceeding the critical threshold. Most mutations are replication errors. For every 109 nucleic base replicates, an error would occur, and so for each cell generation, three mutations occur in each haploid genome.

The mutation rate of autosomal dominant and recessive transmission disorders related to the "X" sex chromosome is less certain and more obvious than the "Y" sex chromosome.

In a genius person, there is an ideal combination of genes, which, however, does not rule out the presence of recessive mutations. Thus, in this context, the sperm could transmit to the offspring not the favorable genes but the recessive mutations, resulting in a disabled child, with neuropsychological pathology, or failure of fertilization expressed by the absence of pregnancy.

All this proves that "genius" is not hereditary, even if there were some exceptions such as Bach, Strauss or Darwin. Last but not least, there are risks of altering the genome of the harvested sperm, such as changes and failures in thawing the collected biological product or failure in fertilization.

"Each of us has, from the beginning, his biological individuality, his uniqueness, because, genetically, no one is identical with anyone, except the monozygotic twins." (Rostand J.)

Conclusions

The intellectual genius is not transmitted as such from parents to descendants, its appearance having as substrate one of the genes of the DNA of the two parents; therefore, the two sex cells and not just one of the two gametes (male and female) determine the genetic support for the procreation of offspring (note that in humans, there is no parthenogenesis).

Through heredity, the child cannot fully possess the biological and cognitive values of the parents, so all the qualities of the sperm donor.

The hereditary transmission and the selection of some exceptional qualities from a parent can be explained by the existence of a mutation, respectively the persistence of the transmission of some values within a family.

Extremely high intellectual capacity and creativity are determined during the nine months of gestation, the gifted child being born with this peculiarity, which, paradoxically, does not exist in his parents. The precocity of the cognitive value of the human brain is proved by the early manifestation, at a very young age, of prodigy children such as Gauss (mathematician), Ampère (physicist), Mozart and Haydn (musicians).

If the genius is "formed" as such, as there is no specific gene, the functional features of the brain in intellectually gifted children could be the consequence of

one or more events in the human genome, namely:

- a genic mutation that affects the structure of DNA (by changing the genetic code)

- a chromosomal mutation that affects the links between genes that are moved from one chromosome to another

- an ideal combination of favorable genes that have removed recessive mutations

- mutations accumulated in the genetic reservoir occurred as a replication error

- mutations caused by external factors (temperature, etc.) or internal (interaction between genes and hormones) that influence the intrauterine development of the product of conception.

According to Andre Lwoff, "the body is a specific, complex and organized system, endowed with genetic continuity, through reproduction." However, genius is **NOT a direct hereditary inheritance of the intellectual qualities of one or both parents. Intelligence, which is said to have a strong genetic transmission, may be due to a favorable mutation in the genome.**

Conflict of interest

The authors have no conflict of interest to make the declaration, had full access to all the data in the study and takes responsibility for the accuracy of the data analysis.

References

1. Abeliovich D, Carmi R, Karplus M, Bar-Ziv J, Cohen M M: Monosomy 21: A possible stepwise evolution of the karyotype. AJMG 4:279-286, 1979.
2. C. Arseni, Stoica I., Tudor I.: Crizele epileptice - Diagnostic electroclinic, fiziopatologie si tratatment, Ed. Scrisul Romanesc, 1978
3. Bareliuc Lucia., Neagu Natalia : Embriologie umană normală și patologică; Ed.Medicală p.13-32, 1987
4. Boue J., Boue A.: Anomalies de la structure cromosomique et arrêts du développment. In: Génétique médicale. Acquisitions et perspectives, Flammarion Médicale, Paris,1981, p.113.
5. Burgoyne P.: The origine of men with two X chromosome, Nature, 1984, p.109, 307.
6. Chandely AC (1981b) Male infertility and meiosis in man. In: Frajese G, Hafez EFE, Conti C, Fabbrini A (eds) Oligozoospermia: Recent progress in andrology. Raven Press, New York, pp 247–265
7. Emery A.E.H. et colab.: Prospective study of genetic counselling, Brit. Med. J., 1979,1, 1 253.
8. Fraser Roberst J. A., Pembrey M.E.: An introduction to medical genetics, Oxford Univ. Press, New York-Toronto, 1979.
9. Frezal J.: La carte et la génome. In: Génetique médicale. Acquisitions et perspectives, Flammarion Médicale, Paris,1981, p.229.
10. Galton F.: Heredity, talent and character, Mac Millan's Magazine, London, 1865, p.12
11. Gedda L., Brenci G.: Twin studies and chronogenetics, Progr. Clin. Biol. Rev., 198169C, 169.
12. Hager T.: High risk „heart" families: a genealogical look, J. Amer. Med. Ass.,1983,250, 13, 1, 663.
13. Kretschmer Ernst: Körperbau und Charakter: Untersuchungen zum Konstitutionsproblem und zur Lehre von den Temperamenten, 1921.
14. Howell R.R.: Genetic disease: the present status of treatment. In:Medical Genetics (Mc Kusick V.A., Clairbone R. -Editors), H.P. Publ. C., New York, 1973, p.271
15. Jacob F., Monod J., Lwoff A : Genetic regulatory mechanisms in the synthesis of proteins. J. Mol. Biol. 3: 318–356, 1961..
16. Leder PH.: Mechanisms of gene evolution, J. Amer. Med. Ass., 1982, 248, 13, 1 582.
17. Maximilian C., Ioan Doina: Genetica medicală, Ed.Medicală 1986, p.11-36.
18. Maximilian C.: Aventura geneticii, Ed.Albatros,

Buc, 1978, p.33
19. Muller H. J.: Studies in Genetics: The Selected Papers of H. J. Muller, Indiana University Press, 1962.
20. Muller H.J.: Modern Concept of Nature, ISBN 0-87395-096-8, SUNY Press, 1973.
21. Novitski E.: Human genetics, The Macmillan Company, New York, 1982.
22. Optz J.M., Shapiro S.S., Vehling D.T.: Genetic causes and workup of male and female infertility. Postgrad. Med. 1979, 65, 5, 247.
23. Rostand J. : Ecrits sur l' hérédité , Seghers, Paris, p.168, 1968.
24. Rothenberg, Albert FLIGHT FROM WONDER: An Investigation of Scientific Creativity. Oxford University Press, 2015
25. Slater E., Cowie V.: The genetics of mental disorders. Oxford Univ. Press, Londra, 1971.
26. Stern C.: Principals of human genetics, vol. III, Ed. W.H. Freeman, San Francisco, 1973.
27. Turner G., Opitz J.M.: Editorial comment: X-linkage mental retardation, Amer. J. Med. Genet., 1980, 7, 407.
28. Vogel F.: Mutation in man. In: Principles and practice of medical genetics (Emery A.E.H., Rimoin D.L.- Editors), Ch. Livingstone Co., Edinburgh, 1983.
29. Hugo de Vries: Die Mutationstheorie („Teoria mutațiilor"- The Mutation Theory) 1901 – 1903.