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CAUSES OF PURINE AND PYRIMIDINE METABOLISM DISORDERS

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Annotation: Purines and pyrimidines are essential building blocks of DNA, RNA, and compounds involved in cellular energy transfer and biosynthetic reactions (e.g., adenosine triphosphate, ATP). Purine and pyrimidine disorders have a wide spectrum of signs and symptoms, including autism, kidney stones, susceptibility to infections, and severe intellectual disability. Symptoms may present from infancy to old age. Most metabolic screening tests do not detect disorders of purine or pyrimidine metabolism; hence, they must be specifically sought out by having specialized analyses performed. Adenosine deaminase (ADA) deficiency results in the accumulation of 2′-deoxyadenosine in the circulating white blood cells (lymphocytes). This, in turn, causes a decreased number of lymphocytes and a drastically increased susceptibility to infection (severe combined immunodeficiency, SCID). Bone marrow transplantation may be curative, and gene therapy has shown promise, but enzyme replacement therapy is the standard treatment.

Key words: Adenosine,ADA,metabolic syndrome,phosphoribosyltransferase, purine,pyrimidine.

Lesch-Nyhan syndrome is an X-linked condition caused by a deficiency in the enzyme hypoxanthine-guanine phosphoribosyltransferase. The nervous system is affected, resulting in writhing movements in the first year of life, after a period of normal development. A particularly troublesome feature is the occurrence of self-mutilation. Intellectual disability is also common. Most individuals with Lesch-Nyhan syndrome excrete a large amount of uric acid in their urine, leading to gout, kidney stones, and possible kidney failure. A high fluid intake and the drug allopurinol are helpful in treating the joint and kidney problems, but have no effect on the severe intellectual disability. Physical restraint and extraction of the teeth are the only successful therapies for the self-injurious behaviour.

Purine, any of a class of organic compounds of the heterocyclic series characterized by a two-ringed structure composed of carbon and nitrogen atoms. The simplest of the purine family is purine itself, a compound with a molecular formula $C_5H_4N_4$. Purine is not common, but the purine structure occurs in many natural substances.

Uric acid, the first purine derivative to be discovered, was isolated in 1776 from urinary calculi; xanthine was obtained from the same source in 1817. Xanthine also occurs in tea, as does caffeine, another purine compound. Guanine, found in guano, the accumulated excrement and dead bodies of birds, bats, and seals, and adenine were identified in 1891 as products of the chemical decomposition of nucleic acids, the cell constituents that determine hereditary characteristics. Purine itself and several purine compounds were synthetically prepared from uric acid in the 1890s.

Pyrimidine, any of a class of organic compounds of the heterocyclic series characterized by a ring structure composed of four carbon atoms and two nitrogen atoms. The simplest member of the family is pyrimidine itself, with molecular formula $C_4H_4N_2$.

Several pyrimidine compounds were isolated between 1837 and 1864, but their structures were not recognized until 1868. Some well-known pyrimidine compounds include cytosine, thymine, and uracil, present in nucleic acids; thiamine (vitamin B_1); and sulfadiazine, sulfamerazine, and sulfamethazine, drugs used in therapy of bacterial and viral diseases.

Energy transfer, the change of energy from one form to another. According to the first law of thermodynamics, energy can be neither created nor destroyed; in other words, the total energy of the universe remains constant. For example, when a block slides down a slope, the potential energy of the block sitting at the top of the slope is converted to the kinetic energy of the block's motion. When friction slows the block to a stop, the kinetic energy is converted to thermal energy. Energy is not created or destroyed but merely changes forms, being transferred from potential to kinetic to thermal energy. *Excitation*, in physics, the addition of a discrete amount of energy (called excitation energy) to a system—such as an atomic nucleus, an atom, or a molecule—that results in its alteration, ordinarily from the condition of lowest energy (ground state) to one of higher energy (excited state).

In nuclear, atomic, and molecular systems, the excited states are not continuously distributed but have only certain discrete energy values. Thus, external energy (excitation energy) can be absorbed only in correspondingly discrete amounts.

Thus, in a hydrogen atom (composed of an orbiting electron bound to a nucleus of one proton), an excitation energy of 10.2 electron volts is required to promote the electron from its ground state to the first excited state. A different excitation energy (12.1 electron volts) is needed to raise the electron from its ground state to the second excited state. Similarly, the protons and neutrons in atomic nuclei constitute a system that can be raised to discrete higher energy levels by supplying appropriate excitation energies. Nuclear excitation energies are roughly 1,000,000 times greater than atomic excitation energies. For the nucleus of lead-206, as an example, the excitation energy of the first excited state is 0.80 million electron volts and of the second excited state 1.18 million electron volts.

The excitation energy stored in excited atoms and nuclei is radiated usually as visible light from atoms and as gamma radiation from nuclei as they return to their ground states. This energy can also be lost by collision.

The process of excitation is one of the major means by which matter absorbs pulses of electromagnetic energy (photons), such as light, and by which it is heated or ionized by the impact of charged particles, such as electrons and alpha particles. In atoms, the excitation energy is absorbed by the orbiting electrons that are raised to higher distinct energy levels. In atomic nuclei, the energy is absorbed by protons and neutrons that are transferred to excited states. In a molecule, the energy is absorbed not only by the electrons, which are excited to higher energy levels, but also by the whole molecule, which is excited to discrete modes of vibration and rotation.

Energy level, in physics, any discrete value from a set of values of total energy for a subatomic particle confined by a force to a limited space or for a system of such particles, such as an atom or a nucleus. A particular hydrogen atom, for example, may exist in any of several configurations, each having a different energy. These energy levels, in their essentials, remain fixed and are referred to as stationary states.

The energy level of a hydrogen atom, or any submicroscopic system, may change from one configuration to another, however, by emitting or absorbing a discrete amount of energy. The atom, or system, is said to undergo a transition between two energy levels when it emits or absorbs energy. The lowest energy level of a system is called its ground state; higher energy levels are called excited states. See also Franck-Hertz experiment.

Mechanics, science concerned with the motion of bodies under the action of forces, including the special case in which a body remains at rest. Of first concern in the problem of motion are the forces that bodies exert on one another. This leads to the study of such topics as gravity, electricity, and magnetism, according to the nature of the forces involved. Given the forces, one can seek the manner in which bodies move under the action of forces; this is the subject matter of mechanics proper.

Historically, mechanics was among the first of the exact sciences to be developed. Its internal beauty as a mathematical discipline and its early remarkable success in accounting in quantitative detail for the motions of the Moon, Earth, and other planetary bodies had enormous influence on philosophical thought and provided impetus for the systematic development of science.

Mechanics may be divided into three branches: statics, which deals with forces acting on and in a body at rest; kinematics, which describes the possible motions of a body or system of bodies; and kinetics, which attempts to explain or predict the motion that will occur in a given situation. Alternatively, mechanics may be divided according to the kind of system studied. The simplest mechanical system is the particle, defined as a body so small that its shape and internal structure are of no consequence in the given problem. More complicated is the motion of a system of two or more particles that exert forces on one another and possibly undergo forces exerted by bodies outside of the system.

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