Spinal muscular atrophy (SMA)

Spinal muscular atrophy (SMA) is a genetic disease affecting the part of the nervous system that controls voluntary muscle movement. SMA is *muscular* because its primary effect is on muscles that don't receive signals from these nerve cells. *Atrophy* is the medical term for getting smaller, which is what generally happens to muscles when they're not active. It is a one of the most common genetic conditions affecting children. It is estimated that one in every 6,000 to 10,000 babies worldwide is born with SMA.

In the most common form of SMA (chromosome 5 SMA, or SMN-related SMA), there is wide variability in age of onset, symptoms and rate of progression. In order to account for these differences, the chromosome 5 SMA often is classified into types 1 through 4Research has focused on strategies to increase the body's production of SMN protein, lacking in the chromosome 5-related forms of the disease. Approaches in this and other forms of SMA include methods to help motor neurons survive in adverse circumstances.

The U.S. Food and Drug Administration on Dec. 23, 2016, approved nusinersen (brand name Spinraza) for the treatment of SMA. Spinraza is designed to treat the underlying defect in SMA, which means it potentially may be effective at slowing, stopping or perhaps reversing the symptoms of SMA.

Congenital Adrenal Hyperplasia, CAH

The term congenital adrenal hyperplasia (CAH) encompasses a group of autosomal recessive disorders, each of which involves a deficiency of an enzyme involved in the synthesis of cortisol, aldosterone, or both. Deficiency of 21-hydroxylase, resulting from mutations or deletions of *CYP21A*, is the most common form of CAH, accounting for more than 90% of cases. Approximately two-thirds of people with classic CAH are classified as having the salt-losing form, while one-third have the simple-virilizing form. In both forms, affected females have genital ambiguity. It is estimated that 1 in 18,000 children are born with congenital adrenal hyperplasia.

A child with classic CAH may experience:

- A lack in the production of cortisol in both the salt-losing and simple-virilizing forms

- A lack in the production of aldosterone in the salt-losing form
- Excess production of the male sex hormones (androgens such as testosterone

Hereditary Hearing Loss and Deafness during puberty

Approximately 1 in 1,000 children suffers by severe hearing loss during pre-school puberty. Hereditary hearing loss and deafness can be regarded as syndromic or non-syndromic. Syndromic hearing impairment is associated with malformations of the external ear, with malformations in other organs, or with medical problems involving other organ systems. Nonsyndromic hearing impairment has no associated visible abnormalities of the external ear or any related medical problems; however, it can be associated with abnormalities of the middle ear and/or inner ear. Approximately 80% of prelingual deafness is genetic, most often autosomal recessive and nonsyndromic. The most common cause of severe-to-profound autosomal recessive nonsyndromic hearing loss in most populations is mutation of *GJB2*. The most common cause of mild-to-moderate autosomal recessive hearing loss is mutation of *STRC*. The 35delG mutation of the GJB2 gene is founded in raised levels in Greece, where the frequency of carriers are 3.5% of the population.

Deficiency of Biotinidase

Biotinidase deficiency is an inherited disorder in which the body is unable to recycle the vitamin biotin (Vitavin B7). If untreated, young children with profound biotinidase deficiency usually exhibit neurologic abnormalities including seizures, hypotonia, ataxia, developmental delay, vision problems, hearing loss, and cutaneous abnormalities (e.g., alopecia, skin rash, and candidiasis). Older children and adolescents with profound biotinidase deficiency often exhibit motor limb weakness, spastic paresis, and decreased visual acuity. Once vision problems, hearing loss, and developmental delay occur, they are usually irreversible, even with biotin therapy. Individuals with partial biotinidase deficiency may have hypotonia, skin rash, and hair loss, particularly during times of stress.

Thalassemia

Thalassemia is a blood disorder passed down through families (inherited) in which the body makes an abnormal form of hemoglobin. Hemoglobin is the protein in red blood cells that carries oxygen. The disorder results in large numbers of red blood cells being destroyed, which leads to anemia. Thalassemia is the most common inherited diseases since there is no effective treatment for thalassemia, the most appropriate way of dealing with prevention is prognosis.

Hemochromatosis

Hemochromatosis is a disease in which too much iron builds up in your body. Diagnosis usually occurs during adulthood, with symptoms including fatigue or skin pigmentation or even the appearance of cardiac, hepatic, endocrine and musculoskeletal complications.

Galactosemia

Galactosemia is a disorder that affects how the body processes a simple sugar called galactose. Galactosemia is a metabolic disease with symptoms such as lethargy, vomiting, diarrhea, growth failure and jaundice. However, the above symptoms do not allow the immediate diagnosis of the disease as there are relatively common. Molecular prenatal testing allows pathogenic mutations to be identified in parents.

G6PD Enzyme Deficiency

G6PD deficiency is an inherited condition in which the body doesn't have enough of the enzyme glucose-6-phosphate dehydrogenase, or G6PD, which helps red blood cells (RBCs) function normally. This deficiency can cause **hemolytic anemia**, usually after exposure to certain medications, foods, or even infections. In Greece, it is estimated that 5% of the population suffers by lack of enzyme, with men appeared with manifesting symptoms of the disease more frequently than women.

Thromboembolic disease

Thromboembolic disease is one of the major causes of morbidity and mortality in the whole population. Pregnancy is considered to be as a high risk for the onset of symptoms of this disease. Molecular control for thrombophilia is usually performed at the beginning of pregnancy in order to be treated any thromboembolic incident immediately. In addition, certain thrombophilia genotypes have been associated with miscarriages as well as preeclampsia.