Answers to picture quiz

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Answer 1

Caveolinopathy - limb girdle muscular dystrophy type 1C

Caveolin-3 is a protein that in humans is encoded by the CAV3 gene. Mutations identified in this gene lead to interference with protein oligomerization or intra-cellular routing, disrupting caveolae formation and resulting in Limb-Girdle muscular dystrophy type-1C (LGMD-1C), HyperCKemia, distal myopathy or rippling muscle disease (RMD). Other mutations in caveolin causes Long QT Syndrome or familial hypertrophic cardiomyopathy, although the role of Cav3 in Long QT syndrome has recently been disputed.

Answer 2

Emery-Dreifuss muscular dystrophy (EDMD)

EDMD is characterized by the clinical triad of (1) joint contractures that begin in early childhood; (2) slowly progressive muscle weakness and wasting initially in a humero-peroneal distribution and later extending to the scapular and pelvic girdle muscles; and (3) cardiac involvement that may include palpitations, presyncope and syncope, poor exercise tolerance, and congestive heart failure. The X-linked form is caused by mutations in EMD, the gene encoding emerin; the dominant/recessive forms are caused by mutations in LMNA, the gene encoding lamin A/C.

Answer 3

McArdle disease

Glycogen storage disease type V (GSD-V) is a metabolic disorder, more specifically a glycogen storage disease, caused by a deficiency of myophosphorylase. Its incidence is reported as 1 in 100,000 approximately the same as glycogen storage disease type I. GSD type V is also known as McArdle disease or muscle phosphorylase (myophosphorylase) deficiency. The disease was first reported in 1951 by Dr. Brian McArdle of Guy's Hospital, London.

The onset of this disease is usually noticed in childhood but often not diagnosed until the third or fourth decade of life. Symptoms include exercise intolerance with muscle pain, early fatigue, painful cramps, and myoglobin in the urine (often provoked by a bout of exercise).

Patients may exhibit a ''second wind'' phenomenon. This is characterized by the patient's better tolerance for aerobic exercise such as walking and cycling after approximately 10 minutes. This is attributed to the combination of increased blood flow and the ability of the body to find alternative sources of energy, like fatty acids and proteins. In the long term, patients may exhibit renal failure due to the myoglobinuria, and with age, patients may exhibit progressively increasing weakness and substantial muscle loss. Muscle biopsy showss subsarcolemmal cytoplasmic vacuoles (Blebs).

Answer 4

Inclusion body myositis

Inclusion body myositis (IBM) is an inflammatory muscle disease characterized by slowly progressive weakness and wasting of both distal and proximal muscles, most apparent in the muscles of the arms and legs. There are two types: sporadic inclusion body myositis (sIBM), which is more common, and hereditary inclusion body myopathy (hIBM). This is the commonest form of acquired myopathy in people over 50 years.

The muscles in the thighs – the quadriceps and the muscles in the arms that control finger flexion-making a fist-are usually affected early on. Common early symptoms include frequent tripping and falling, weakness going up stairs and trouble manipulating the fingers (including difficutly with tasks such as turning doorknobs or gripping keys). Dysphagia is present in from 40 to 85% of IBM cases.

Answer 5

Kennedy disease

Also known as spinal and bulbar muscular atrophy (SBMA), spinobulbar muscular atrophy, bulbo-spinal atrophy, X-linked bulbospinal neuropathy (XBSN), X-linked spinal muscular atrophy type 1 (SMAX1). Kennedy's disease is a debilitating neurodegenerative disorder resulting in muscle cramps and progressive weakness due to degeneration of motor neurons in the brain stem and spinal cord.

The condition is associated with mutation of the androgen receptor (AR) gene and is inherited in an X-linked recessive manner. The androgen receptor gene that is mutated in SBMA is located on the X chromosome, and the effects of the mutation may be androgen-dependent, thus only males are fully affected. Females are rarely affected; female carriers tend to have a relatively mild expression of the disease if they show symptoms at all.

Because of its endocrine manifestations related to the impairment of the AR gene, SBMA can be viewed as a variation of the disorders of the androgen insensitivity syndrome (AIS). Endocrine manifestations are gynaecomastia, impotence, erectile dysfunction, reduced fertility, low sperm count, testicular atrophy.