5 Ablepharon Macrostomia Syndrome

Synonym: AMS

Ablepharon macrostomia syndrome (AMS) is an extremely rare inherited disorder characterized by various physical abnormalities affecting the head and facial (craniofacial) area, the skin, the fingers, and the genitals. In addition, afflicted individuals may have malformations of the nipples and the abdominal wall. Infants and children with AMS may also experience delays in language development and, in some cases, mental retardation. In infants with ablepharon macrostomia syndrome, characteristic craniofacial features may include absence or severe underdevelopment of the upper and lower eyelids (ablepharon or microblepharon) as well as absence of eyelashes and eyebrows; an unusually wide, “fish-like” mouth (macrostomia); and/or incompletely developed (rudimentary), low-set ears (pinnae). Abnormalities of the eyes may occur due to, or in association with, ablepharon or microblepharon. Individuals with AMS may also have additional characteristic features including abnormally sparse, thin hair, thin, wrinkled skin with excess (redundant) folds; webbed fingers with limited extension; and/or malformations of the external genitals. In some cases, additional features associated with AMS may include absent or abnormally small (hypoplastic) nipples and/or abdominal wall abnormalities. Although the exact cause of ablepharon macrostomia syndrome is not fully understood, some cases suggest that the disorder may be inherited as an autosomal recessive genetic trait.

The following organizations may provide additional information and support:

- Centers for Disease Control and Prevention
- NIH/National Institute of Allergy and Infectious Diseases
- World Health Organization (WHO) Regional Office for the Americas (AMRO)

6 Acanthocheilonemiasis

Acanthocheilonemiasis is a rare tropical infectious disease caused by a parasite known as Acanthocheilonema perstans, which belongs to a group of parasitic diseases known as filarial diseases (nematode). This parasite is found, for the most part, in Africa. Symptoms of infection may include red, itchy skin (pruritus), abdominal and chest pain, muscular pain (myalgia), and areas of localized swelling (edema). In addition, the liver and spleen may become abnormally enlarged (hepatosplenomegaly). Laboratory testing may also reveal abnormally elevated levels of certain specialized white blood cells (eosinophilia). The parasite is transmitted through the bite of small flies (A. culicoides).

The following organizations may provide additional information and support:

- Rainbow Research Group
- NIH/National Eye Institute
- NIH/National Institute of Diabetes, Digestive & Kidney Diseases
- Retinitis Pigmentosa International
- Kidney Diseases
- NIH/National Institute of Arthritis and Musculoskeletal and Skin Diseases

7 Acanthocytosis

Synonyms: Bernstein-Forziari Syndrome
Low-Density Beta Lipoprotein Deficiency

Acanthocytosis is a digestive disorder that is characterized by the absence of very low density lipoproteins (VLDL) and chylomicrons in the plasma. Chylomicrons are very small fatty droplets that are covered with a beta-lipoprotein and perform an essential function in fat transport in the blood and, thus, in fat metabolism. The absence of VLDL and of chylomicrons interferes with the absorption of fat and leads to excessive fats excretion (steatorrhea). Other symptoms include abnormal red blood cells (acanthocytes), a vision disorder (retinitis pigmentosa), and impaired muscle coordination (ataxia).

The following organizations may provide additional information and support:

- CLMB (Children Living with Inherited Metabolic Diseases)
- Foundation Fighting Blindness, Inc.
- National Tay-Sachs and Allied Diseases Association, Inc.
- NIH/National Institute of Diabetes, Digestive & Kidney Diseases
- Retinitis Pigmentosa International

8 Acanthosis Nigricans

Synonym: AN

Disorder Subdivisions
- Acanthosis Nigricans with Insulin Resistance Type A
- Acanthosis Nigricans with Insulin Resistance Type B
- Benign Acanthosis Nigricans
- Drug-Induced Acanthosis Nigricans
- Hereditary Benign Acanthosis Nigricans
- Malignant Acanthosis Nigricans
- Pseudoacanthosis Nigricans

Acanthosis nigricans (AN) is a skin disorder characterized by abnormally increased coloration (hyperpigmentation) and “velvety” thickening (hyperkeratosis) of the skin, particularly of skinfold regions, such as of the neck and groin and under the arms (axillae). Various benign (non-cancerous) forms of AN have been identified in which the disorder may be inherited as a primary condition or associated with various underlying syndromes; an excess accumulation of body fat (obesity); or the use of certain medications (i.e., drug-induced AN). In other instances, AN may occur in association with an underlying cancerous tumor (i.e., malignant AN). Experts suggest that AN may be a skin manifestation of insulin resistance, which is a condition characterized by impaired biological responses to insulin. Insulin, a hormone produced by the pancreas, regulates blood glucose levels by promoting the movement of glucose into cells for energy production or into the liver and fat cells for storage. Glucose is a simple sugar that is the body’s primary source of energy for cell metabolism. Insulin resistance may be associated with various disorders, including obesity and non-insulin-dependent type II diabetes mellitus. In individuals with type II diabetes mellitus, the pancreas produces insulin but the body becomes resistant to its effects, leading to insufficient absorption of glucose and abnormally increased glucose levels in the blood (hyperglycemia) and urine. As a result, there may be a gradual onset of certain symptoms, including excessive urination (polyuria) and increased thirst (polydipsia), and the development of particular complications without appropriate treatment.

The following organizations may provide additional information and support:

- March of Dimes Birth Defects Foundation
- NIH/National Institute of Arthritis and Musculoskeletal and Skin Diseases

9 Achalasia

Synonyms: Cardiospasm
Dysynergia Esophagus
Esophageal Aperistalsis
Megaeosophagus

Achalasia is a rare disorder of the esophagus characterized by the abnormal enlargement of the esophageal wall, impairment of the ability of the esophagus to push food down toward the stomach (peristalsis), and the failure of the ring-shaped muscle (sphincter) at the bottom of the esophagus to relax.

The following organizations may provide additional information and support:

- March of Dimes Birth Defects Foundation
- NIH/National Digestive Diseases Information Clearinghouse

10 Achard-Thiers Syndrome

Synonym: Diabetic Bearded Woman Syndrome

Achard-Thiers syndrome is a rare disorder that occurs primarily in postmenopausal women and is characterized by type 2 (insulin-resistant) diabetes mellitus and signs of androgen excess. The exact cause of this syndrome is unknown.

The following organizations may provide additional information and support:

- American Diabetes Association
- National Adrenal Diseases Foundation
- NIH/National Institute of Diabetes, Digestive & Kidney Diseases

11 Achondrogenesis

Synonyms: Chondrogenesis Imperfecta
Hypochondrogenesis
Lethal Neonatal Dwarfism
Lethal Osteochondrodysplasia
Neonatal Dwarfism

Disorder Subdivisions
- Achondrogenesis, Type III
- Achondrogenesis, Type IV
- Fraccaro Type, Achondrogenesis (Type II)
- Houston-Harris Type, Achondrogenesis (Type IA)
- Langer-Saldino Type, Achondrogenesis (Type II)

Achondrogenesis is a very rare disorder characterized by extreme short-limbed dwarfism, lack of