Cantu Syndrome

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Clinical characteristics:

Cantú syndrome is characterized by congenital hypertrichosis; distinctive coarse facial features (including broad nasal bridge, wide mouth with full lips and macroglossia); enlarged heart with enhanced systolic function or pericardial effusion and in many, a large patent ductus arteriosus (PDA) requiring repair; and skeletal abnormalities (thickening of the calvaria, broad ribs, scoliosis, and flaring of the metaphyses). Other cardiovascular abnormalities may include dilated aortic root and ascending aorta with rare aortic aneurysm, tortuous vascularity involving brain and retinal vasculature, and pulmonary arteriovenous communications. Generalized edema (which may be present at birth) spontaneously resolves; peripheral edema of the lower extremities (and sometimes arms and hands) may develop at adolescence. Developmental delays are common, but intellect is typically normal; behavioral problems can include attention-deficit/hyperactivity disorder, autism spectrum disorder, obsessive-compulsive disorder, anxiety, and depression.

Diagnosis/testing:

The diagnosis of Cantú syndrome is established in a proband with suggestive clinical findings and a heterozygous pathogenic variant in

ABCC9

or

KCNJ8

identified by molecular genetic testing. Some individuals with a clinical diagnosis of Cantú syndrome have not had a pathogenic variant identified in either gene, suggesting the existence of another as-yet unidentified causative gene.

Management:

Treatment of manifestations:

Surgical or device closure of PDA in infancy or early childhood as needed. Pericardiocentesis and pericardial stripping as needed to treat pericardial effusion. Compression stockings for peripheral edema; shaving and (in teenagers and adults) use of depilatories or laser hair removal for hypertrichosis; bracing and/or surgery as needed for scoliosis; individualized management for migraine headaches and developmental delays if present.

Surveillance:

Yearly echocardiogram and electrocardiogram to monitor cardiac size and function, as well as for evidence of pericardial effusion. Clinical evaluation and cardiac biomarkers to monitor late development of high-output cardiac failure. Monitor for evidence of peripheral edema annually starting in adolescence and for scoliosis with physical examination, followed by spine radiographs as needed. Monitor for a history of persistent headaches or other neurologic symptoms, which may require brain imaging for cerebral vasculature abnormality and evaluation by a neurologist.

Evaluation of relatives at risk:

If the pathogenic variant in an affected family member is known, relatives at risk who are suspected of having Cantú syndrome can be offered molecular genetic testing to clarify their genetic status. Family members who are affected should be evaluated and monitored for cardiac manifestations, scoliosis, and peripheral edema.

Genetic counseling:

Cantú syndrome is inherited in an autosomal dominant manner. Each child of an individual with Cantú syndrome has a 50% chance of inheriting the pathogenic variant and being affected. Prenatal and preimplantation genetic testing are possible if the pathogenic variant has been identified in an affected family member.