CollegeNews
AMERICAN COLLEGE OF MEDICAL GENETICS

Announcements

Costello Syndrome

Costello Syndrome is a rare genetic disorder, with about 150 people identified worldwide since it was first recognized as a distinct syndrome in 1971. It is suspected to be the result of an autosomal dominant gene. Individuals with Costello Syndrome typically exhibit 3 clinical phases: (1) polyhydramnios during pregnancy, and increased birth weight; (2) "coarse" facial features and hypotonia that may lead to a suspicion of a storage disorder in infancy through early childhood. There is loose skin, lax joints, dramatic weight loss, failure to thrive, global developmental delay, and general medical fragility. (3) In late childhood, nutrition improves, and the children are generally more medically stable. Distinctive skin findings include papillomata, hyperkeratosis, and pigmentation. Other problems include heart anomalies (a broad spectrum from mild arrhythmia to severe hypertrophic cardiomyopathy), malignant tumors (especially rhabdomyosarcoma), and orthopedic issues. Efforts are being made in the US and UK to understand the clinical features of Costello syndrome, particularly its relationship to cancer.

At present, diagnosis is clinical. Storage disorders must be ruled out. Children diagnosed with Noonan and Cardio-Facio-Cutaneous Syndrome have been rediagnosed with Costello Syndrome, and vice-versa. Although there are similarities to other syndromes, Costello syndrome has a distinctive appearance, which can be recognized in our children's faces. Both the CostelloKids Web site and the brochure have multiple images of the children.

If you suspect one of your patients may have Costello Syndrome, we have several ways to help. We have a Web site, http://www.costellokids.org.uk/; or you can contact the Costello Program at the duPont Hospital for Children, http://www.nemours.org/no/aidhc/svcs/div2626.html. Brochures are available and can be ordered through the CostelloKids Web site. We welcome your participation.

Kidney disease

Kidney disease? Call NIH at 1-800-411-1222 for information on new kidney studies, including Lupus Nephritis, Membranous Nephropathy, and Focal Segmental Glomerulosclerosis. Treatment provided at no cost. Transportation may be provided. (TTY: 1-866-411-1010) E-mail prpl@cc.nih.gov. NIH is part of Department of Health and Human Services.

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Diplomates Certified In 2002–2003

ABMG is proud to acknowledge the individuals who achieved certification in 2002–2003:

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