


[My Profile](#) [Log In](#)

Home / Life Sciences / Genetics




American Journal of Medical Genetics Part A [e-mail](#) [print](#)

[What is RSS?](#)

American Journal of Medical Genetics Part A
Early View (Articles online in advance of print)

Published Online: 30 Oct 2007
 Copyright © 2007 Wiley-Liss, Inc.

[Save Title to My Profile](#)
[Set E-Mail Alert](#)


 Go to the homepage for this journal to access trials, sample copies, editorial and author information, news, and more. ▶

SEARCH All Content
 Publication Titles

[Advanced Search](#)
[CrossRef / Google Search](#)
[Acronym Finder](#)


SEARCH IN THIS TITLE

American Journal of Medical Genetics Part A

SEARCH BY CITATION


Vol: Issue: Page:

INDEXED BY



This journal is listed in the U.S. National Library of Medicine's PubMed index.

REPRINT INQUIRIES






Need a reprint?

Paper or electronic reprints are available for all content published on Wiley InterScience. Inquiries can be submitted online.

[Find out more about reprints](#)

LATEST NEWS & INFORMATION

Winners of the 6th Annual Wiley Prize in Biomedical Sciences

Congratulations to:
Dr. F. Ulrich Hartl
 and

[Save Article to My Profile](#)
[Download Citation](#)
[Previous Abstract](#)
[Next Abstract](#)

Abstract | [References](#) | Full Text: [HTML](#) | [Related Articles](#) | [Citation Tracking](#)

Clinical Report

The Hunter-MacDonald syndrome with expanded phenotype including risk of meningioma: An update and review[†]

Linlea Armstrong^{1*}, Gail E. Graham², R. Neil Schimke³, Debra L. Collins⁴, Daniel J. Kirse⁵, Fiona Costello⁶, Holly H. Ardinger⁷

¹Department of Medical Genetics, University of British Columbia, Vancouver, British Columbia, Canada
²Department of Pediatrics, University of Ottawa, Ottawa, Ontario, Canada
³Departments of Medicine and Pediatrics, Kansas University Medical Center, Kansas City, Kansas
⁴Department of Medicine, Kansas University Medical Center, Kansas City, Kansas
⁵Department of Otolaryngology-Head and Neck Surgery, Wake Forest University, Winston-Salem, North Carolina
⁶Department of Neurosurgery, University of Ottawa, Ottawa, Ontario, Canada
⁷Section of Medical Genetics and Molecular Medicine, Children's Mercy Hospitals and Clinics, Kansas City, Missouri

email: Linlea Armstrong (llarmstrong@cw.bc.ca)

*Correspondence to Linlea Armstrong, Provincial medical genetics programme, The Women's and Children's Health Centre of British Columbia, C234-4500 Oak Street, Vancouver BC V6H 3N1, Canada.

[†]How to cite this article: Armstrong L, Graham GE, Schimke RN, Collins DL, Kirse DJ, Costello F, Ardinger HH. 2007. The Hunter-MacDonald syndrome with expanded phenotype including risk of meningioma: An update and review. *Am J Med Genet Part A*.

KEYWORDS

multiple epiphyseal dysplasia • familial meningioma • low frequency hearing loss • scoliosis • camptodactyly • thumb subluxation

ABSTRACT

Hunter-MacDonald syndrome (HMS) is a rare, autosomal dominant skeletal dysplasia with multiple malformations. The skeletal manifestations of HMS include short stature, scoliosis, epiphyseal dysplasia with early osteoarthritis leading to joint replacement, prominent humeral insertions for the deltoids, camptodactyly, subluxation of the thumbs, and malformed feet. Craniofacial manifestations include normal head circumference, tall forehead, bitemporal narrowing, ptosis, short palpebral fissures, and short philtrum. Decreased hearing acuity, transient cranial nerve palsies, congenital heart defects, and meningioma are also reported. Herein, we present two cases, and, through review of the manifestations of HMS in affected and at-risk family members, we have observed that predisposition to brain tumor is a cardinal feature of this condition. © 2007 Wiley-Liss, Inc.

Received: 5 December 2006; Accepted: 11 June 2007

DIGITAL OBJECT IDENTIFIER (DOI)

10.1002/ajmg.a.31998 [About DOI](#)