Chiari I malformation and neurofibromatosis type 1.

Submitted by abartol on Tue, 10/08/2013 - 11:41am

Title
Chiari I malformation and neurofibromatosis type 1.

Publication Type
Journal Article

Year of Publication
2004

Authors
Tubbs, Shane, S Rutledge, L Kosentka, A Bartolucci, AA Oakes, WJerry

Journal
Pediatr Neurol

Volume
30

Issue
4

Pagination
278-80

Date Published
2004 Apr

ISSN
0887-8994

Keywords
Adolescent, Adult, Arnold-Chiari Malformation, Child, Child, Preschool, Cranial Fossa, Posterior Decompression, Surgical, Female, Humans, Infant, Male, Mesoderm, Neurofibromatosis 1, Neurologic Examination, Pregnancy, Retrospective Studies

Abstract
Single case reports exist in the medical literature of patients with tonsillar ectopia, i.e., the Chiari I malformation and neurofibromatosis type 1. However, large series of patients with either of these entities have not been examined for the presence of both defects. We have retrospectively examined two large groups of pediatric patients: Group I, with the primary diagnosis of Chiari I malformation, who have undergone posterior fossa decompression for symptomatology; and Group II patients, who have been observed in our hospital's neurofibromatosis clinic for evaluation. Of 130 surgically addressed Chiari I malformations (Group I), we determined that 5.4% of these patients had the additional diagnosis of neurofibromatosis type 1. Of Group II patients (198) who underwent imaging of the brain, 8.6% were found to have a concomitant Chiari I malformation. These data suggest that Chiari I malformation and neurofibromatosis type 1 are not spurious findings but rather true associations. We hypothesize that the same early dysgenesis of mesoderm that is widely accepted as a culprit in the genesis of many Chiari I malformations is the same pathology affecting primitive development of tissues involved in many patients with neurofibromatosis type 1. Perhaps these data will aid in the determination of a genetic locus for the Chiari I malformation.

DOI
10.1016/j.pediatrneurol.2003.09.013

Alternate Journal

PubMed ID
15087107