

Central nervous system malformations in Mohr's syndrome

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SUMMARY A boy with severe developmental delay, bilateral, symmetrical hallucal duplication, and accessory alveolar frenula was found to have radiological evidence of a large arachnoid cyst compressing the cerebellum and brain stem. We review neurological abnormalities in Mohr's syndrome.

The oro-facial-digital syndrome (OFD) type II, or Mohr's syndrome, has been designated, as a separate entity from OFD type I, on the basis of autosomal recessive inheritance, bilateral hallucal reduplication, normal hair and skin, and the occasional presence of a conductive hearing loss.¹⁻³ There has been a small number of reports of neurological features in association with Mohr's syndrome, but in general patients with OFD type II differ from type I in that intelligence is usually normal and central nervous system malformations are unusual.^{4,5} We report a

case with features of Mohr's syndrome in a patient who had a large posterior fossa subarachnoid cyst and occipital meningocele, which have not previously been described in this condition.

Case report and investigations

The proband, a boy, was the fourth child born to healthy, non-consanguineous, Irish parents. The pregnancy was remarkable only in that the mother noticed reduced fetal movements throughout. Spontaneous vaginal delivery took place at 38 weeks. Birth weight was 3400 g. He was noted to be hypotonic and clinical suspicion of a meningocele was confirmed radiologically. He was a poor feeder and early in life developed marked apnoeic episodes. These were particularly prone to occur when hungry or otherwise aggravated and were often associated with cyanosis. Marked hypotonia followed these episodes but no seizures were noted. The meningocele was excised at the age of three months.

His developmental milestones were markedly



FIG 1 Accessory alveolar frenula.

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P J Roberts Southampton *Developmental Aspects of Craniofacial Dysmorphology* Michael Melnick, Ronald Jorgenson, 1979
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 American Journal of Medical Genetics ,1983 **Scanning Electron Microscopy** ,1986 Vols for 1968 77 include the proceedings of the annual Scanning Electron Microscope Symposium sponsored by the IIT Research Institute and other workshops Human Embryology and Developmental Biology E-Book Bruce M. Carlson, 2008-11-25 This thoroughly revised 4th edition offers both clear descriptions and explanations of human embryonic development based on all the most up to date scientific discoveries and understanding Particular attention is paid to the fundamental aspects of molecular mechanisms in development introducing you to major families of important developmental molecules Clinical aspects of development are covered throughout in boxed sections of text First rate illustrations complete this essential package Integrates contemporary developmental knowledge with classical embryological understanding Interprets complex molecular developments to help you learn how exactly the embryo develops Presents first rate clinical photos and clear drawings to help you to memorize and understand normal and abnormal development Uses clear sections within the chapter and summaries at the end of each to help you navigate this complex subject Includes review questions at the end of each chapter to help you assess your knowledge Provides more coverage of molecular development to help you interpret complex information Revises the section on the development of the head particularly useful for dental students

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Table of Contents Recent Advances In The Developmental Biology Of Central Nervous System Malformations

1. Understanding the eBook Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - The Rise of Digital Reading Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Advantages of eBooks Over Traditional Books
2. Identifying Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - User-Friendly Interface

4. Exploring eBook Recommendations from Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Personalized Recommendations
 - Recent Advances In The Developmental Biology Of Central Nervous System Malformations User Reviews and Ratings
 - Recent Advances In The Developmental Biology Of Central Nervous System Malformations and Bestseller Lists
5. Accessing Recent Advances In The Developmental Biology Of Central Nervous System Malformations Free and Paid eBooks
 - Recent Advances In The Developmental Biology Of Central Nervous System Malformations Public Domain eBooks
 - Recent Advances In The Developmental Biology Of Central Nervous System Malformations eBook Subscription Services
 - Recent Advances In The Developmental Biology Of Central Nervous System Malformations Budget-Friendly Options
6. Navigating Recent Advances In The Developmental Biology Of Central Nervous System Malformations eBook Formats
 - ePub, PDF, MOBI, and More
 - Recent Advances In The Developmental Biology Of Central Nervous System Malformations Compatibility with Devices
 - Recent Advances In The Developmental Biology Of Central Nervous System Malformations Enhanced eBook Features
7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Highlighting and Note-Taking Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Interactive Elements Recent Advances In The Developmental Biology Of Central Nervous System Malformations
8. Staying Engaged with Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Recent Advances In The Developmental Biology Of Central Nervous System Malformations

9. Balancing eBooks and Physical Books Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Recent Advances In The Developmental Biology Of Central Nervous System Malformations
10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
11. Cultivating a Reading Routine Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Setting Reading Goals Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Carving Out Dedicated Reading Time
12. Sourcing Reliable Information of Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Fact-Checking eBook Content of Recent Advances In The Developmental Biology Of Central Nervous System Malformations
 - Distinguishing Credible Sources
13. Promoting Lifelong Learning
 - Utilizing eBooks for Skill Development
 - Exploring Educational eBooks
14. Embracing eBook Trends
 - Integration of Multimedia Elements
 - Interactive and Gamified eBooks

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