PRACTICE PARAMETER: EVALUATION OF THE CHILD WITH MICROCEPHALY (AN EVIDENCE-BASED REVIEW)

This is a summary of the American Academy of Neurology (AAN) and Child Neurology Society guideline (*Neurology*® 2009;73:887–897) regarding evaluation of the child with microcephaly. Recommendations are presented for neuroimaging, genetic testing, and screening for coexistent conditions.

Please refer to the full guideline at www.aan.com for more information, including additional tables and appendices, and the AAN's definitions of the levels of recommendations and classifications of evidence.

Microcephaly (head circumference [HC] more than 2 standard deviations [SDs] below the mean for age and gender) may result from any insult that disturbs early brain growth and can be seen in association with hundreds of genetic syndromes. Annually, approximately 25,000 infants in the United States will be diagnosed with microcephaly (HC <-2 SD).

What is the role of diagnostic testing of children with microcephaly?

NEUROIMAGING	
Weak evidence	Neuroimaging may be considered useful in identifying structural causes in the evaluation of the child with microcephaly (**Level C**).
Clinical context*	MRI often reveals findings that are more difficult to visualize on CT, such as migrational disorders, callosal malformations, structural abnormalities in the posterior fossa, and disorders of myelination, and is considered the superior diagnostic test.

GENETIC TESTING	
Weak evidence	Targeted genetic testing may be considered in the evaluation of the child with microcephaly in order to determine a specific etiology (**Level C**).
Clinical context*	Microcephaly has been associated with numerous genetic etiologies. Because the genetics of microcephaly is a rapidly evolving field, current data underestimate the importance and relevance of genetic testing as part of the diagnostic evaluation. Many of the microcephaly genes have been associated with specific phenotypes, allowing targeted clinical testing. However, insufficient data showing the diagnostic yield of these tests preclude specific recommendations for use.

METABOLIC TESTING	
Insufficient evidence	There is insufficient evidence to support or refute obtaining metabolic testing on a routine basis for the evaluation of the newborn or infant with microcephaly (**Level U**).
Clinical context*	Microcephaly is common in global developmental delay (GDD) and the yield of metabolic testing may be higher when the following are present: parental history of consanguinity, family history of similar symptoms in relatives, episodic symptoms, developmental regression, extracranial organ failure, or specific findings on neuroimaging. Metabolic testing may have a higher yield when microcephaly remains unexplained after other evaluations have been done.

What neurological disorders are associated with microcephaly?

Four common neurologic disorders frequently occur in children with microcephaly: epilepsy, cerebral palsy, developmental and learning disorders, and ophthalmological and audiological disorders. Severe forms of epilepsy also can be seen in subpopulations. These are associated with genetic disorders (see Table 2 in the published guideline).

EPILEPSY	
Weak evidence	Because children with microcephaly are at risk for epilepsy, physicians may consider educating caregivers of children with microcephaly on how to recognize clinical seizures (**Level C**).
Insufficient evidence	There are insufficient data to support or refute obtaining a routine EEG in a child with microcephaly (**Level U**).

CEREBRAL PALSY	
Strong evidence	Because children with cerebral palsy (CP) are at risk for developing acquired microcephaly, serial HC measurements should be followed (**Level A**).
Weak evidence	Because children with microcephaly are at risk for CP, physicians and other care providers may consider monitoring them for early signs so that supportive treatments can be initiated (**Level C**).

MENTAL RETARDATION	
Strong evidence	Because children with microcephaly are at risk for developmental disability, physicians should periodically assess development and academic achievement to determine whether further testing and rehabilitative efforts are warranted (**Level A**).

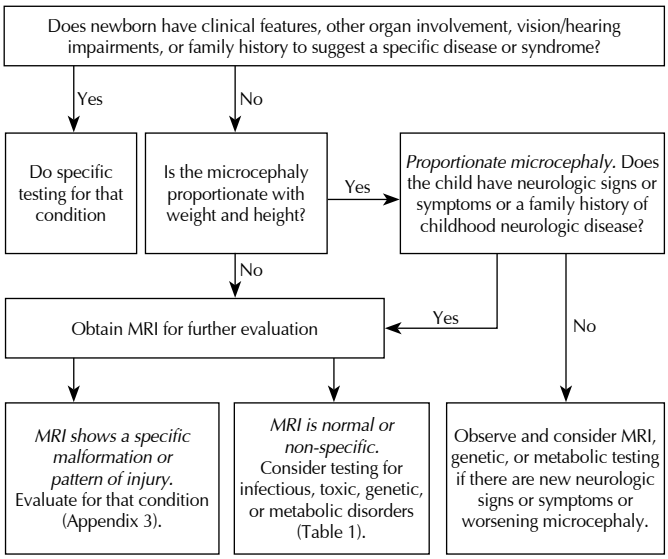
OPHTHALMOLOGICAL AND AUDIOLOGICAL DISORDERS	
Weak evidence	Screening for ophthalmological abnormalities in children with microcephaly may be considered (**Level C**).
Clinical context*	Certain microcephaly syndromes are characterized by sensory impairments. Early identification of visual and hearing deficits may help identify a syndrome and the need for supportive care of the child.

Clinical Context*

Congenital Microcephaly

Many medical experts advocate doing a prompt, comprehensive evaluation of congenital microcephaly, given the risk of neurodevelopmental impairment and the parental anxiety associated with the diagnosis. Consulting a neurologist and geneticist can help to guide the diagnostic evaluation and support and educate families. Establishing a more specific diagnosis provides valuable information regarding etiology, prognosis, treatment, and recurrence risk. The initial history, examination, and screening laboratory testing may suggest a specific diagnosis or diagnostic category, allowing further screening or testing to be targeted. If the initial evaluation is negative and the child appears to have isolated microcephaly, a head MRI may help to categorize the type of microcephaly.

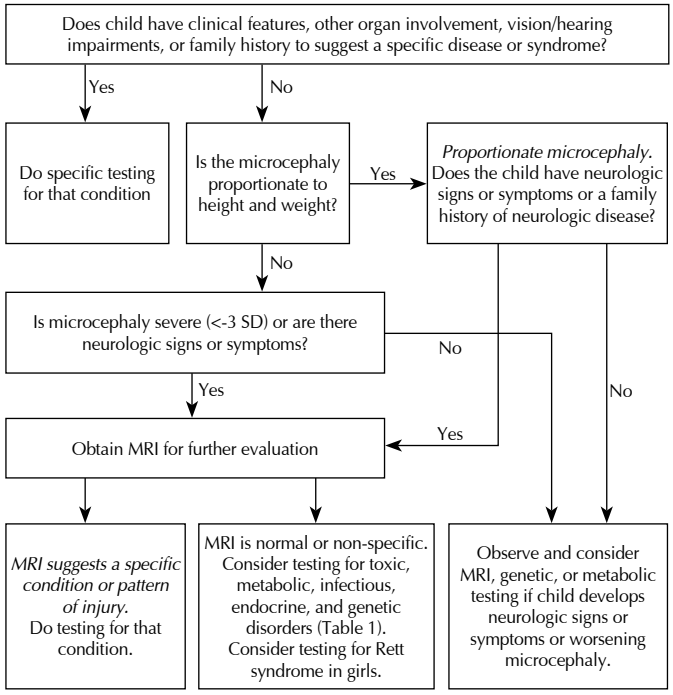
Figure 1: Evaluation of congenital microcephaly



Postnatal Onset Microcephaly

Microcephaly from acquired insults to the CNS or from progressive metabolic/genetic disorders is usually apparent by age 2 years. Mild or proportionate microcephaly may go unrecognized unless a child's HC is measured accurately. Making comparisons to parents' HCs may be important as familial forms of mild microcephaly have been described. Currently available assessment tools may not ultimately establish a specific etiologic diagnosis.

Figure 2: Evaluation of postnatal onset microcephaly



*Clinical context slightly abridged. See the published guideline for the complete text.

CHILD NEUROLOGY SOCIETY

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