**Consultation with *the Specialist* : Ataxia**

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# Consultation with the Specialist

## Ataxia

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A child or adolescent who has ataxia should stimulate a pediatrician to consider a broad array of possible diagnoses. The incoordination of muscle action can be caused by pathology located anywhere from the cerebral cortex to the involved muscle. Ataxia can be frightening to all involved, and the tendency is to call a neurologist quickly. However, without the information gathered from a careful history and physical examination, unnecessary expensive tests might be ordered and consultation with the neurologist would not be efficient.

The Table provides an extensive but not exhaustive list of possible causes of ataxia. However, the omitted diagnoses are extremely rare and most likely would be made only by a subspecialist after an exhaustive evaluation. The most common causes of ataxia generally can be diagnosed and managed by the general pediatrician. They fall in the categories of acute infectious, postinfectious, and metabolic etiologies. The diagnoses that are life-threatening if not diagnosed and treated rapidly must be considered first, and they also fall primarily in the infectious and metabolic categories. These include acute bacterial infections and metabolic aberrations, such as hypoglycemia, hyponatremia, hyperammonemia, and ingestion of alcohol and other drugs and toxins. Neoplasias and vascular problems are less common but also can fall in this category; they generally require management by a subspecialist.

If the atactic child is stable and in no acute distress, the general pediatrician should proceed with a careful history, physical examination, and basic diagnostic tests before consulting a subspecialist.

### History

A careful history involving the timing of onset and nature of the ataxia can provide invaluable information. Has the child had a previous episode of ataxia? Are there associated signs and symptoms, such as fever, nausea, vomiting, lethargy, headache, and head tilting?Has there been a recent viral infection or cough? (Varicella is probably the most common infectious cause of ataxia.) Has the child or

**TABLE. Causes of Ataxia**

**Idiopathic (Postinfectious) Acute Cerebellar Ataxia**
1. Enteroviruses
2. Influenza viruses
3. Mycoplasmal organism
4. Myxovirus
5. Varicella

**Infections**
1. Bacterial meningitis
2. Viral meningoencephalitis

**Metabolic Disorders**
1. Biotinidase deficiency
2. Dominant hereditary ataxias
3. Drugs/poisons/toxins
4. Hartnup disease
5. Hyperammonemia
6. Hypoglycemia
7. Hyponatremia
8. Lactic-pyruvic acidemia
9. Organic acidemia
10. Porphyria
11. Refsum disease
12. Vitamin E deficiency
13. Wernicke encephalopathy
14. Wilson disease

**Vestibular Disturbances**
1. Antibiotic toxicity to labyrinth
2. Endolymphatic hydrops
   - Congenital infection
   - Inner ear malformation
   - Meniere disease (idiopathic)
   - Posttraumatic event
3. Labyrinthitis (infectious or postinfectious)
4. Perilymphatic fistula (posttraumatic or postinfectious)

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youth been taking medications or might he or she have ingested a drug or poisons? Did the teenager drink alcohol or take other drugs? Could he or she possibly have attempted suicide? Does the child have or is there a family history of sickle cell disease, metabolic diseases, epilepsy, migraines, or Friedreich ataxia? Has there been a recent change in mental status?

## Physical Examination

The neurologic portion of the physical examination should be complete but reserved for last to lessen the likelihood that important general findings are missed. Important general findings include scars or crusty varicella lesions on the skin, the presence of fluid in the middle ear, cervical adenopathy, chest rales, cardiac arrhythmias, murmurs, splenomegaly, hepatomegaly, abdominal masses, and decreased muscle strength. As part of the neurologic examination, papilledema or hemorrhages in the fundi should be noted. Also, whenever possible, asynergia (elicted by excessive wrist extension on attempted finger flexion), dysmetria (error in estimating the movement distance of the examiner's finger as it is moved suddenly to various locations in one place), intention tremor, and smoothness of completing a complex movement should be evaluated. Obviously, some of these latter evaluations would be extremely difficult in a small child.

## Diagnostic Tests

One of the more common causes of ataxia can be evaluated by performing a dextrostix test. Other readily available, relatively inexpensive, and low-risk diagnostic tests are measuring serum electrolyte levels, including determining bicarbonate level, screening for toxins, and performing a lumbar puncture (if the diagnosis of meningitis is entertained and the fundi are normal).

More elaborate and expensive tests, such as computed tomography or magnetic resonance imaging, are performed best after consultation with a neurologist.

**TABLE. continued**

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| **Neoplasms** |
| 1. Neuroblastoma |
| 2. Tumors of brain stem/cerebellum |
| **Vascular Disorders** |
| 1. Arteriovenous malformation |
| 2. Basilar artery migraine |
| 3. Cerebellar hematoma/hemorrhage |
| 4. Cervical vertebral fracture or dislocation |
| 5. Coagulopathy |
| 6. Homocystinuria |
| 7. Infarction |
| 8. Sickle cell crisis |
| 9. Stroke |
| 10. Vasculitis |
| 11. Ataxia telangiectasia |
| **Weakness** |
| 1. Guillain-Barré syndrome |
| 2. Myasthenic syndrome |
| 3. Tick paralysis |
| 4. Transverse myelitis |
| **Other Causes** |
| 1. Epilepsy |
| 2. Friedreich ataxia |
| 3. Hydrocephalus |
| 4. Hypothyroidism |

**IN BRIEF**

# Craniosynostosis

**Craniosynostosis: A Review of 519 Surgical Patients.** Shillito J Jr, Matson DD. *Pediatrics.* 1968;41:829–853

**Craniosynostosis Update 1987.** Cohen MM Jr. *Am J Med Genet Suppl.* 1988;4:99–148

**Perspectives on Craniosynostosis.** In: Cohen MM Jr, ed. *Craniosynostosis: Diagnosis, Evaluation, and Management.* New York, NY: Raven Press; 1986:21–57

**Syndromes, Genetics, and the Craniofacial Complex.** Marion RW. In: Goodrich JT, Hall CD, eds. *Craniofacial Anomalies: Embryology to Surgical Management.* New York, NY: Thieme Med Publishers, in press.

On the surface, premature closure of one or more sutures of the skull may appear to be a straightforward malformation that leads to a specific, recognizable pattern of symptoms and signs, an anomaly that can be corrected simply by a surgical procedure. But in fact, craniosynostosis, which occurs in approximately 1 in 2500 children, is a dynamic process, the external reflection of a series of complex conditions that may cause an underlying abnormality not only in the skull but in other systems.

In most cases, surgical repair of both the fused sutures and the resulting craniofacial asymmetry will resolve the primary problems caused by craniosynostosis, namely, compression of the brain because of lack of space, functional anomalies of the eye due to aberrant placement of the orbits, and sometimes, grotesque distortion of the facies. However, in many other instances, the affected child will be left after surgery with ongoing, unresolved medical and surgical problems. It is the task of the pediatrician to identify these latter individuals prior to surgery.

Members of the family often are the first to notice that the infant's head is asymmetric. This realization may occur in the immediate newborn period or develop gradually over the course of the first few months of the infant's life. Usually, the parents bring their concern to the pediatrician, who, on physical examination, may appreciate the child's unusual head shape, the lack of mobility at the suture lines, or the presence of bony ridging. Such children require further evaluation.

The first step in the evaluation involves obtaining radiographs of the skull in the anteroposterior and lateral planes. Although these films may

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