Recognizing Problems That Require Further Evaluation in Children with NF1

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This month, I'd like to address an issue that often arises in the minds of parents whose children have been newly diagnosed with NF1. These parents often ask when they should be concerned about an issue or symptom that they notice in their child. I don't think parents should assume the task of being their child's doctor and become hypervigilant about every potential issue. Instead, parents have the important role and responsibility of nurturing and caring for their child. However, it's natural for parents to experience anxiety about possible complications of NF1, and we do want parents to be alert to any potentially serious complication that may develop. The key is in separating everyday aches and pains from important symptoms, and the central question becomes: What are the complications that, if detected early, would allow for better outcomes for children with NF1?

Optic Glioma

A tumor of the optic pathway, or optic glioma, occurs in approximately 15% of children with NF1. These tumors usually occur early in life, between the ages of 18 to 24 months. While more than half of children with optic glioma have no symptoms, some children experience vision loss, usually between the ages of 2 to 6 years. Because very young children don't complain of vision loss, the early presentation of these problems can be subtle. Some signs of possible visual impairment include: tripping over objects or having difficultly navigating physical obstacles; becoming fearful of walking down stairs; and holding objects closer than normal or sitting closer to a screen, such as a television or computer. While we recommend yearly eye exams for children with NF1, parents who recognize these possible signs of vision loss should make an appointment for an evaluation with an NF specialist or pediatric ophthalmologist.

Physical Growth

A physical feature that is common for children with NF1 is that head size tends to be larger than average. However, a sign of concern would be if the size of the head crossed percentile lines as it grew or became noticeably larger in a relatively short period of time. Also, vomiting and lethargy could be a sign of obstructive hydrocephalus, a condition of increased brain fluid pressure that is rare, but more common in people with NF1 and usually occurs in childhood or young adulthood.

Also regarding physical growth, some degree of short stature is common among children with NF1. Slow weight gain is also common, although falling off the growth curve or crossing percentile lines are a cause for concern that requires further evaluation. In some cases, a brain stem tumor or optic glioma can affect the functioning of the hypothalamus where appetite is controlled, resulting in weight loss.

Plexiform Neurofibromas

These tumors, which occur deep in the body and involve large branches of multiple nerves, are usually noticed in the first year of life. They appear as a painless soft tissue swelling of the arm, leg, or around one or both eyes or on the face. Plexiform neurofibromas are believed to be congenital in most cases, although they are not easy to see at birth. Swelling of the upper eyelid in the early years of life could be a sign of a plexiform neurofibroma around the eye, which can grow rapidly in childhood and cause significant disfigurement and interference with vision. Enlargement of an arm or leg can also be an early sign of plexiform neurofibroma.

Bone Dysplasia

This problem is an abnormality of a long bone, usually involving the tibia in the leg but also sometimes affecting the fibula as well as bones in the arms. Bone dysplasia sometimes presents as bowing of a leg in infancy, although this can be difficult to detect early because most infants have some normal leg bowing. By the time a child can stand, one can usually determine if dysplasia is present. An X-ray is performed to confirm the problem, and the child is referred to an orthopedist for treatment with a leg brace to prevent fracture. If a fracture does occur, it can be difficult to treat, which makes early detection of this problem important.

Developmental and Cognitive Issues

Some children with NF1 exhibit low muscle tone, which results in muscles that are less firm and seem weaker than normal. This problem tends to improve over time, but it may evolve into some degree of poor coordination in adolescence and adulthood. Also, learning problems are present in approximately 50% of children with NF1, although this issue may not become apparent until the child has reached school age. Children with NF1 may exhibit problems in maintaining attention, hyperactive behavior, and social immaturity. In some, speech articulation may be affected. Sudden onset developmental delay is not common in children with NF1. If a child is failing to reach developmental milestones or displays signs of learning or cognitive problems, this is a cause for concern and further evaluation.

Headache

I have mentioned the occurrence of headaches in children with NF1 in previous blogs. Most typically these occur intermittently and may be associated with nausea, stomach aches, and vomiting. These signs are suggestive of migraine, which seems to be more common in children with NF1 than in the general population. Another cause of headaches in children with NF1 is Chiari malformation, in which the base of the brain extends below the foramen magnum, which is the space in the skull where the spinal cord connects to the brainstem. This is also more common in children with NF1 than in the general population. Many parents of children with NF1 and headaches worry that the headaches could be a sign of brain tumor. For a brain tumor to cause headaches it requires that the tumor cause increased fluid pressure in the brain. If this does happen, the headaches are usually severe, may wake the child from sleep, and are associated with severe vomiting. A careful physical exam would reveal increased pressure on the optic nerve visible in an eye exam, and would be followed up with an MRI scan. Fortunately, I find that this is an uncommon cause of headaches in children with NF1.