

# STAR Information Series:

## Medical Issues

**Medical Evaluation.** A thorough medical evaluation may be important for children newly diagnosed with autism. Evaluation typically includes a detailed history of pregnancy and delivery, past and current medical problems, early development, behavioral concerns and family history of health, psychiatric or learning problems. A complete physical examination is conducted to look for any medical problems that might interfere with developmental progress, as well as growth parameters, skin findings or dysmorphic features (unusual facial or extremity findings), which might suggest an associated medical or genetic condition. Neurologic evaluation often reveals subtle differences with muscle tone, gait, and fine motor application skills. Audiologic evaluation is essential to rule out problems with hearing acuity; methods of hearing evaluation include audiometric screen, soundfield testing, and brainstem auditory evoked response testing. Ophthalmologic evaluation may be indicated if there are any concerns about vision.

Because autism is associated with other medical or genetic conditions in approximately 10 to 15 percent of cases, additional laboratory workup may be indicated. These tests may include high resolution chromosome analysis, Fragile X DNA testing, thyroid function tests, lead level, and urine metabolic screen. An imaging study of the brain is not routinely obtained, but may be indicated if there are significant motor problems or focal neurologic findings. Sleep deprived EEG's are generally recommended for children with a history of language regression for the purpose of determining whether there is a characteristic pattern which suggests an acquired epileptiform aphasia of childhood. This disorder, also known as Landau Kleffner syndrome, may present with subclinical seizure activity, marked communication impairments and better developed social skills; treatment options include steroids such as ACTH and anticonvulsants. While medical workup is often negative, it is important to rule out an associated medical or genetic condition.

**Sleep Problems.** Certain medical concerns seem to occur fairly frequently for children with autism. Parents often report problems with sleep patterns, such as difficulty going to sleep, frequent waking during the night, and early morning waking. Few studies have looked specifically at the sleep patterns of children with autism, but prevalence estimates of sleep problems in this population range from 44 to 83 percent. Research to correlate parent reports of sleep difficulties and objective sleep study data is now in the early formulation stage. Behavioral interventions are often the first line treatment for sleep difficulties, but sometimes medication can be useful.

**Eating Problems.** Children with autism often have unusual eating patterns. This frequently takes the form of very narrow food preferences, which seem to be based in part on sensory issues (taste, texture, temperature, etc.). Parents sometimes have concerns about the nutritional adequacy of their child's diet, although the few studies which have looked at this issue have generally reported appropriate nutritional profiles.

Behavioral problems surrounding meals (licking and smelling food, throwing food, spitting food out) may also have to be addressed with specific interventions.

**Gastrointestinal Problems.** Some children with autism have gastrointestinal problems. While it is not clear that children with autism have more problems with digestion than typically developing peers, there have been anecdotal reports of diarrhea, stomach pain, and constipation. Several small studies have reported evidence of inflammatory bowel disease, food allergies, or increased urinary peptides in children with autism. Some of the proposed interventions for children with autism have been based on the theory of a gut-brain connection; that is, it has been hypothesized that the behavioral manifestations of autism and gastrointestinal problems are somehow related.

**Seizures.** As mentioned before, seizures occur in approximately one-third of individuals with autism. Peaks of new onset seizure activity occur in infancy and in adolescence. In infancy, seizures are most likely to infantile spasms, which have a very characteristic EEG pattern and which are often associated with significant developmental delay. In adolescence, the seizures may be of any kind. Seizures need to be appropriately evaluated and treated with antiepileptic medications in order that seizure activity is minimized.

**Immunizations.** Immunizations have been a source of concern for some parents. The possibility of a link between the measles-mumps-rubella (MMR) vaccine and autism has been highly controversial since Dr. Andrew Wakefield of the Royal Free Institute of London initially reported on eight of twelve children with “autistic regression” and ileocolonic nodular hyperplasia (inflammatory bowel disease) whose parents attributed the regression to the MMR vaccine. Subsequent large-scale studies have failed to find any causal association between the vaccine and autism. More recently concerns have been raised that thimerosal, a mercury containing preservative used in certain childhood vaccines such as hepatitis B and diphtheria-tetanus-pertussis, may have played a role in the increasing prevalence of autism. Advocates of this theory point to the similarity in behaviors seen in autism and mercury toxicity. While there is no direct evidence that mercury levels found in thimerosal containing vaccines were neurologically harmful, these preservatives have been removed from vaccines. Concerns about mercury exposure from other sources such as fish and dental amalgams remain and have lead some parents to pursue chelation therapy to remove heavy metals. It is clear that further study is needed to determine whether environmental factors such as mercury exposure could have a role in causing autism.

**Biologic Interventions.** Most professionals agree that educational and behavioral interventions constitute the primary treatment for autism. However, biologic interventions have also been proposed. Among the most popular of these is the casein-gluten free diet based on the hypothesis that children with autism have difficulty breaking down proteins found in milk and wheat products. As a result, the peptides from partial breakdown build up in the bloodstream and act as natural opioids, causing behavioral manifestations characteristic of autism. While no controlled studies are available on the efficacy of the casein and gluten free diet, many anecdotal reports from parents have described behavioral improvements and/or improvement in gastrointestinal function following its implementation.

**Vitamins and Supplements.** Vitamin supplements have also been advocated in the treatment of autism. Dr. Bernard Rimland of the Autism research Institute has been a strong proponent of vitamin B6 and magnesium supplements. He cites several studies supporting the efficacy of vitamin B6 in promoting attention and eye contact. Other researchers have been less enthusiastic and refute claims of benefits; they also note the possibility of a peripheral neuropathy as a side effect of excessive vitamin B6. One small study indicated some reduction in stereotypies with vitamin C use, but this finding has not been replicated. There has been recent interest in use of Omega 3 fatty acids because of their role in immune function and neurotransmission, but their use in autism has not been studied. Dr. Mary Megson has proposed use of vitamin A supplements in the recommended daily allowance as a way to boost the immune system and protect against immune insults. It is important to note that large doses of vitamin A can be harmful and may result in increased intracranial pressure. An initial report of improvement in expressive language lead to interest in the use of DMG (dimethylglycine) in autism; however, a subsequent double-blind controlled study yielded no benefit of DMG over placebo.

**Antiyeast Therapy.** Antiyeast therapy has sometimes been utilized based on the theory that children with autism may have an overgrowth of yeast in their intestinal tracts secondary to an immune abnormality or excessive antibiotic use. Anecdotal reports from parents have claimed behavioral benefits in some cases, but no controlled studies of antifungal therapy have been performed.

**Medication.** Medication is sometimes used as an adjunct to educational/behavioral strategies. While there are no medications that change core symptoms of autism, medications may help with behaviors that interfere with learning, social interaction or independent daily living. These behaviors include hyperactivity, self injury, aggression, stereotypies, excessive irritability or anxiety, and sleep disorders. Medication should not be used as a substitute for an appropriate behavioral plan and likely will not be as effective without concomitant behavior strategies in place. It is important that medication options as well as potential benefits and side effects, be discussed with the individual's physician. Families should be aware of risk versus benefit for any proposed medication. If a trial of medication is pursued, close monitoring with feedback from teachers and family members will aid the physician with medication adjustment.

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STAR is an intervention program developed specifically for children and adolescents with autism spectrum disorders (ASDs) and their families. STAR is a program within the Weisskopf Center for the Evaluation of Children (WCEC), a division of the University of Louisville School of Medicine Department of Pediatrics.

