

Tests that help uncover answers to the question- Why is my child behaving this way?

One of the sad facts, when a child is given a diagnosis of “autism spectrum”, is that many of the concerns that parents have about their child, whether it be gastrointestinal, behavioural or physical, are dismissed as “autistic traits”.

It is not uncommon, when we finally determine the underlying pathology or biochemical abnormality that is contributing to the child's symptoms, that the parents, often the mother says *“I just knew that something was very wrong, but no one would listen.”* In one very extreme instance, after an initial evaluation, I insisted that a 6 year old Asperger child should be seen by a neurologist. This mother, to her credit, went through her child's paediatrician, and three other doctors (*one doctor suggested anti-anxiety and antidepressant medication for the mother as she was an “overanxious mother”*), before she was finally able to get a referral. Once the MRI was performed the child was diagnosed with a life **threatening condition** and spent the next 6 weeks in hospital being treated. The mother said *“I knew something was wrong when she was 3 years old”*. **Interestingly, once treated, her Asperger traits disappeared!**

Every child should have a thorough assessment medically, nutritionally, and biochemically to identify barriers that may be preventing them from achieving their full potential.

Traditional Pathology and Integrative Medicine Tests

We are all familiar with the pathology tests that mainstream doctors order. Integrative medicine goes beyond normal pathology tests. These tests have often been developed and are in use in research facilities for years before being made available to mainstream health professionals by specialist laboratory services. They are often slowly picked up and offered by mainstream pathology like folate (MTHFR) gene testing, gene probes for parasites, homocysteine, etc. Included in this article are a selected range of tests that have been shown to be valuable in assessing ASD children. Tests are a useful guide to the practitioner to determine deficiencies or errors in metabolism that may be present in their child and how best to correct these deficiencies. It is not suggested that all tests be done. The child's clinical history will determine which tests will be most useful. Having a blood draw done for a child can be a traumatic experience, however hair, urine and stool tests can be equally useful diagnostic tools.

Pathology Testing - Blood work

Having blood work done gives a good baseline for a child's current health status, as well future trends in their overall health. These can be done on their own, or if blood is being drawn for other testing (e.g. fragile X chromosomal studies), these can be easily added on.

FBE (Full Blood Examination), iron studies, vitamin B12, folate, vitamin D, plasma zinc, serum copper, thyroid function, liver function.

Please note that pathology companies do not use specific age related reference ranges in children for all tests. Therefore your child's test may appear “normal” on the adult range, but be outside of normal range on their age appropriate reference range. Most doctors are not aware of this.

Changing the
way we think
about treating
autism.

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Abdominal (KUB) X-Ray

Essential to determine the degree of **faecal impaction or constipation** in your child and also the location, whether in the upper or lower bowel. Although some doctors are reluctant to order a KUB X-ray, it is not unusual for them to be surprised at the level of stool that is backed up in the child's bowel.

This test requires a referral from your doctor.

Hair Mineral and Tissue Analysis

A hair mineral analysis is useful to determine baseline minerals in the body (calcium, magnesium, iron, selenium, etc) as well as heavy metals (mercury, lead, cadmium, arsenic, etc). A high heavy metal result can be followed up with a blood test to see if there is ongoing exposure to such metals. Low zinc, iron, calcium, magnesium, and others, can also be checked with a blood test.

Urinary Iodine

Iodine deficiency is common in children in Australia. Iodine is essential for normal brain function, including the development of speech and cognitive skills. Recent studies of children in Tasmania and NSW confirms children in general are not consuming sufficient iodine.

Recurrent Iodine Deficiency in Tasmania, Australia: A Salutory Lesson in Sustainable Iodine Prophylaxis and Its Monitoring. Kamala Guttikonda, et al. J Clin Endocrinol Metab, June 2002, 87 (6):2809–2815



Comprehensive Digestive Stool Analysis (CDSA)

If there are unresolved gastrointestinal issues, diarrhoea, loose stools, constipation, abdominal pain and bloating. A Comprehensive Digestive Stool Analysis is superior to the stool testing provided by conventional pathology laboratories. There are different stool tests available, some are based on culturing the micro-organisms, others use DNA technology to detect them. Which one to order depends on what you suspect may be the problem in the gastrointestinal tract. Apart from detecting beneficial, imbalanced and pathogenic bacteria in the bowel, a Comprehensive Digestive Stool Analysis gives valuable additional information, including:

- parasitic infection
- yeast infection
- digestive (pancreatic) function
- absorption ability of fats and carbohydrates
- gut inflammation, including specific markers of inflammation (lysozyme), mucus
- short chain fatty acids - end products of bacterial digestion essential for a healthy bowel and to protect the gut from pathogens, inflammation and for healing the gut.
- immune status - important as the first line of defense against unwanted organisms
- sensitivity testing - performed on unwanted pathogens (that are able to be grown) to determine which pharmaceutical medications or supplements are effective in controlling these pathogens

MHTFR Gene Test

Methylenetetrahydrofolatereductase (MTHFR) is essential for the conversion of homocysteine to methionine via methylation. A polymorphism of the gene results in defective metabolism of folate and consequently many underlying medical conditions.

Sample required: swab from inside of the mouth

Genetic Testing and Nutrigenomics

Although 30% of our DNA or “genome” is pre-programmed and cannot be changed, 70% of our epigenome (“epi” meaning above) or the layer above our DNA, is controllable. The epigenome is influenced by factors such as environment, nutrition, exercise, etc. Nutrigenomics is the study of how your nutritional status affects your epigenome. Gene testing gives a picture of an individual’s genetic predisposition (strengths and weaknesses) and how best to tailor nutritional, lifestyle and nutritional supplementation for that person. This test is not intended to diagnose disease, rather it provides a suitably qualified health professional with information to make decisions as to which areas of a person’s health need to be focussed on.

Organic Acid Testing

Organic acids are metabolic compounds produced in the body during energy production, detoxification, neurotransmitter breakdown, or intestinal microbial activity. Accumulation of specific organic acids in urine often signals a block or a slowing down of these processes. This may be due to a nutrient deficiency, an inherited enzyme deficit, toxic build-up, or drug effect.

This test provides comprehensive information about how well our body biochemistry is working. The areas assessed include:

- carbohydrate metabolism deficiencies - poor ability to process carbohydrates
- energy production deficiencies - how effectively energy is produced in the body
- nutritional markers, including vitamin C, and vitamins B2, B5, B6, B12, biotin and C, CoQ10, N-acetyl-cysteine, lipoic acid
- oxidative stress markers that can indicate damaged DNA in the body
- detoxification ability - including ability to produce glutathione and remove ammonia from the body
- brain neurotransmitter metabolites to assess for adequate serotonin and dopamine production, and inflammation in the brain
- mitochondrial energy production - whether there may be a mitochondrial dysfunction issue
- oxalate accumulation - high oxalates are present in certain foods, which if not removed, can cause tissue damage
- methylation assessment - extremely important in many biochemical pathways in the body
- intestinal overgrowth markers for bacteria, yeast and clostridia species

Pyrrole Testing

People with depression or other mental health disorders often demonstrate an abnormal production of a group of chemicals called ‘pyrroles’. The excessive pyrroles that are produced by pyroluria patients naturally bind with **zinc** and **vitamin B6**, blocking their receptor sites and causing them to become unavailable for their normal functions, **resulting in a deficiency of zinc and vitamin B6**. Pyroluria may be accompanied by other conditions such as; ADD/ADHD, severe depression, Tourette syndrome, ASD, alcoholism, schizophrenia and bipolar disorder (all of these conditions are different manifestations of pyroluria). Up to 50% of ASD children have pyroluria and there does seem to be a familial or genetic component.

There are many signs and symptoms of pyroluria, some of these include:

- poor morning appetite and/or tendency to skip breakfast
- stretch marks (striae) / pale skin / poor tanning
- knee / joint pain and cold hands or feet
- abdominal tenderness and constipation
- light / sound / odour intolerance
- stress intolerance and emotional lability
- explosive anger / anxiety / withdrawal

Sample required: Urine specimen



Why doesn't my doctor or paediatrician know about these tests?

"Why doesn't my doctor know all of this?" It's because the majority of doctors (paediatricians, gastroenterologists, general practitioners, etc.) do not read medical journals or attend conferences that present leading research on the treatment of ASD children. Doctors do not have the time. They are too busy running their practices.

The overwhelming majority of doctors rely on what they learned in medical school – sometimes 20-30 years ago - and on pharmaceutical sales representatives to keep them "up-to-date" on new drug information. Many studies brought to physicians for "educational purposes" are highly filtered to support their pharmaceutical product.

This concern is particularly clear in an article published in the *New England Journal of Medicine* entitled *Clinical Research to Clinical Practice-Lost in Translation*. The author, Dr. Claude Lenfant, M.D., states there is great concern that doctors continue to rely on what they learned 20 years before and are uninformed about new scientific findings. This article states that very few doctors learn about new discoveries at scientific conferences or from medical journals, with a view to translating this knowledge into enhanced treatments for their patients.

Clinical Research to Clinical Practice — Lost in Translation? Claude Lenfant, M.D. N Engl J Med 2003;349:868-74.