Two Important Laboratory Tests for Difficult Cases
By Kelly Dorfman M.S., L.D.N., Nutritionist and Cofounder DDR

Test 1 – The Smoking Gun for Mercury Toxicity

Most experts agree that mercury and other heavy metals damage the developing nervous system, and that exposure to environmental toxins can contribute to developmental delays. However, proving that a specific poison has harmed a particular child is extremely difficult. One reason is that isolating single factors is elusive when children are in daily contact with a multitude of pesticides, industrial chemicals, and solvents. Even in clear cases of overexposure, such as mercury loading from vaccinations, testing blood levels does not yield answers because the metals are stored in fat tissue, not blood.

At last, labs have the expertise to test blood or urine samples for molecular damage caused by a wide variety of toxins. For girls who are not toilet trained or others whose disabilities make urine collection difficult, plasma blood levels are an option. The tests measure the presence of porphyrins, a chemical ring structure that the body uses to make hemoglobin. It is the "heme" of hemoglobin. Hemoglobin is a porphyrin ring with iron in the middle. Because of the presence of iron, blood, which contains hemoglobin, is red. Chlorophyll, in contrast, is a porphyrin ring, with magnesium is the middle, thus making it green.

Mercury and other toxins interfere with the production of porphyrin at specific places on the ring. The result is malformed or incompletely formed porphyrin that the body excretes because it cannot use it to build hemoglobin. Have you ever noticed that some children with delays appear unusually pale? That's because a defect in hemoglobin production from either inadequate iron or poor porphyrin genesis gives them pasty complexions.

Scientists believe that mercury interferes with porphyrin production at the fifth and sixth step, resulting in excretion of the unfinished porphyrin. The urine test detects specific incomplete, unusable porphyrins discarded before the disrupted step, such as pentacarboxy porphyrin (5-CP) and coproporphyrin (4-CP), which denote the presence of mercury. Other toxins disrupt production at other junctures, resulting in the elevation of different porphyrins.

In the United States, Lab Corp and Quest Laboratories both offer tests that measure porphyrins in blood plasma and urine. While both labs usually take insurance, they have limited experience with the test. Laboratoire Philippe Auguste in Paris has broader experience with the testing and provides clearer results. Send a urine sample by regular air mail and it takes a week to 10 days to process. Contact them at: contact@labbio.net. The cost at this writing is 90 euros (approximately $120) plus shipping.

Test 2 – Avoiding Bad Drug Reactions

Many patients experience one of three types of bad reactions to a medication.

The mode of action can be wrong for the condition. For example, taking a medication for depression that works by raising serotonin when you unknowingly have high serotonin already will cause disassociation.

The mode of action is correct and a secondary problem develops in response to the therapy. Using antibiotics to treat bronchitis and then getting a yeast infection, is an example of this chain reaction.

Side effects result from poor processing and/or excreting of the drug. Incomplete and faulty detoxification of a substance can cause symptoms such as headaches, rashes, paradoxical responses (i.e. over-activity instead of sedation), and emotional swings. Recently, a boy took Celexa for sensory anxiety. Initially, the young man had improved sensory function, was happy and relaxed. Then overnight he became pathologically agitated and distressed because his body could not longer break down the drug.

Now we have a test that pinpoints which patients might have the third type of reaction. This test is especially important for children with autism spectrum disorders, who have weak detoxification systems. Because doctors are relying increasingly on medications as a part of biomedical intervention, they must prescribe medications using the "trial and terror method." Give the drug and watch nervously for bad reactions. The jury is not in yet on the efficacy of this trend, and parents should proceed with caution.

The body breaks down and excretes drugs through a series of steps. First, it identifies a chemical that is unnecessary. Then a group of enzymes called cytochrome P450 enzymes act on the medication or chemical and prepare it for excretion. A different P450 enzyme acts upon each drug to break it down. If a patient inherits a weak gene that creates a specific P450 enzyme, and then takes a drug that needs that enzyme, the drug will break down properly, but toxic by-products and serious side effects will result.

How does the test work? It measures the genes regulating a patient's P450 enzymes. Because pharmaceutical companies are now required to identify which cytochrome P450 enzymes processes each medication, a doctor can choose only medications that match a patient's high functioning detox pathways, thus avoiding side effects.

Several laboratories offer this blood test. The most complete and least costly is from Genova Diagnostics (formerly Great Smokies Diagnostic Lab). At $399 it requires a physician's order. Call 800-522-4762 or go to www.GDX.net.

For youngsters on several medications, GeneMedRx.com, a computerized subscription service collates the known drug detox pathways and genetic information. Since several medications can overwhelm even strong detox pathways if they use the same P450 enzyme, use this resource (free for a short time) to check if multiple medications are overloading a P450 enzyme, even without detox gene information.

The future is here. Exciting new laboratory tests can now tell if mercury is present, even if it is hiding, and prevent drug reactions by testing how an individual's unique detoxification system operates. For children with developmental delays, understanding individual differences in detoxification is critically important.