What are neurotransmitters?

Normal brain function depends on good communication of information between nerve cells. Neurotransmitters are the chemical messengers that work inside the brain to deliver messages between the nerve cells. Nerve cells send out neurotransmitters to activate or inhibit neighbouring nerve cells, speeding up or slowing down nerve functions such as muscle coordination, behavior, mood, blood flow, body temperature and pain.

Some examples of neurotransmitters that work in the brain are:

- Dopamine
- Serotonin
- Norepinephrine
- Epinephrine
- GABA

Neurotransmitters play a vital role in everyday life and functioning.

What is neurotransmitter disease?

Neurotransmitter disease (ND) is an umbrella term for the group of diseases that affect how neurotransmitters are made, transported, or broken down in the brain.

There are many common NDs such as:

- Parkinson’s
- Alzheimer’s
- Depression

Since we are a metabolic clinic, we focus on the diagnosis and treatment of neurometabolic NDs. Examples of these are:

- Dopamine deficiency (ie Dopa-responsive dystonia)
- Pterin deficiencies
- Cerebral folate deficiency
- Serotonin deficiency syndrome
How do people get neurotransmitter disease?

ND are inborn errors of metabolism. They are genetic. This means that the person has the disorder from the time they are conceived*, although the condition might manifest in infancy, childhood or adulthood.

At conception, the baby receives two sets of genetic material, one from the mother, and one from the father. This genetic material, called DNA, acts as a recipe for the baby’s development.

DNA includes information about the baby’s eye and hair colour, gender and even whether the baby will be right or left-handed. DNA comes in units called genes. Each pair of genes gives directions to a certain part of the body.

In most cases**, NDs are caused by getting two copies of the faulty gene, one from the mother and one from the father. This is called autosomal recessive inheritance. If the child inherits only one copy of the gene, they are a carrier for the disorder but are not affected. There is a 1 in 4, or 25% chance that 2 carriers of the gene will have a baby with an ND. Boys and girls are equally affected.

* Cerebral folate deficiency can also be caused by antibodies the body produces against its own folate receptors.
** Dopa-responsive dystonia is an autosomal dominant disorder. This means that a child only needs to inherit one copy of the faulty gene (from either the mother or the father) to be affected by the condition, or it can happen as a new mutation.

How is neurotransmitter disease diagnosed?

When you have unexplained neurological symptoms, the doctor will take some fluid from the space below your spinal cord by doing a lumbar puncture. This procedure is done in the doctor’s office. You will be asked to lie on your side in the fetal position. The doctor will put some freezing in your low back. When the area is numb, the doctor will take some cerebral spinal fluid (CSF)
from your spinal column. You will need to lie flat for an hour after the procedure so that you decrease your risk of developing a headache. The CSF will be analyzed at the lab to determine if you have a neurotransmitter disease. The results take about 4 to 6 weeks.

**What are the symptoms of neurotransmitter disease?**

When neurotransmitters aren’t working as they should, normal brain and spinal cord function is disrupted. People with ND may have some or all of the following symptoms:

- Seizures
- Problems walking
- Problems with mood
- Problems with speech
- Migraine headaches with or without paralysis
- Weakness
- Bladder problems
- Dystonia (twisting, repetitive movements and abnormal postures of body parts) that gets worse in the evening

**How is neurotransmitter disease treated?**

The symptoms of the specific neurotransmitter disease determine treatment. There is no miracle cure for ND. There are, however, different things that may help.

**MEDICATIONS**

The medications that are prescribed depend on the specific ND and symptoms that a person has. Possible medications include:

1. Levo Dopa
2. Carbidopa
3. carnitine
4. 5 hydroxytryptophan (5 HTP)
5. anti seizure medications
6. folinic acid
7. tetrahydrobiopterin (BH 4)
DIET

Although there is no specific diet that has been shown to help improve the symptoms of neurotransmitter disorders, maintaining a healthy diet according to Canada’s food guide will help the person with a neurotransmitter disorder to stay well.

ENERGY CONSERVATION

People with neurotransmitter disease tend to tire easily. It is important for them to pace themselves and to alternate between rest and periods of activity to conserve energy. Sleep is very important. Symptoms may increase when the person is fatigued. Exercise, stress, illness and time of day can cause fatigue.

Regular visits to the metabolic clinic are needed to consult with the team. Referrals can be made to other specialists to closely monitor the effects of the neurotransmitter disease on the body.

How can friends and family help?

Understanding that the person with neurotransmitter disease is not “crazy” or “lazy” is the best help that you can offer. Recognize and accept that they have physical limitations and may tire easily. Become familiar with how their specific disorder affects them. Don’t make assumptions about their level of physical or mental functioning. Remember that the person is feeling frustrated by how quickly their energy level can change. The person may get angry or very depressed and sad. Sometimes the person may be forgetful or have difficulty with planning activities.

Some ideas ...
• Make your offers of help concrete. Instead of saying, “What can I do to help?” or “If there’s anything I can do, let me know”, give clear offers.

Examples are:

“I’ll take care of the kids on Friday night so you and Bob can have a night on your own”

“I’m going to the grocery store tomorrow morning. What can I pick up for you?”

“Why don’t you go and lie down and I’ll vacuum the family room?”

“I’ll take Anne shopping so you can have a break”

Concrete offers like these are easier to accept than offers that the person may perceive as you just “trying to be nice”. Also, there is no decision-making involved, something the person you are trying to help may not have a lot of energy for.

• For people with severe physical limitations from their disorder, offer as much “normal” activity as possible. Include them in activities. Don’t treat them as different. Don’t make assumptions about their capabilities based on visible disabilities.

• For people with less severe limitations from their disorder, remember that their energy level will vary greatly during the day. Learn to look for the telltale signs of fatigue such as slow movement or memory problems. Expect less when fatigue is showing.

• Offer to give caregivers breaks.

• Learn as much as you can about the disorder and help to explain it to others. This not only increases awareness of neurotransmitter disease, it helps the person affected to have a break from explaining it over and over again.
Helpful Resources

The Adult Metabolic Diseases Clinic
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Pediatric Neurotransmitter Disease Association
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