

## ODD MEDICAL DISORDERS

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I write about these because I was reminded of them recently and they are odd enough to be interesting, even if tragic. Most are extremely rare and are due to genetic mutation. This article is meant to engender sympathy and gratitude that no matter how bad you feel, it could be worse. While you may experience some of the common symptoms described, please don't jump to conclusions about having one of these diagnoses and bother your doctor about it, except for those who at high for coronary heart disease, who might ask their doctor for a homocysteine blood test.

**Cotard's syndrome** is a rare neuropsychiatric disorder in which a patient insists that they or their body parts are dead, dying, or don't exist. While there is no known proximate cause, it may occur in people with dementia, encephalopathy, multiple sclerosis, Parkinson's disease or schizophrenia. There is no definitive cure, though Electroconvulsive Therapy (ECT) and antipsychotic, antidepressant and mood stabilizer medications can reduce symptoms.

**Misophonia**, which I have, is a relatively common multi-sensory aversion disorder that causes an exaggerated, negative emotional reaction, ranging from anxiety to rage, that is triggered by repetitive sounds like chewing, slurping, clicking, ticking, dripping, sniffing, tapping or barking. It's frustrating when people making these noises are not sensitive to the discomfort they cause. It is possibly associated with higher cognitive intelligence and/or music or language abilities. According to various studies of the disorder, about ten percent of sufferers also have autism-spectrum, ADHD or obsessive-compulsive disorders, but there is no scientific proof of a common etiology. Treatments include noise-cancelling headphones, cognitive

behavioral therapy and avoidance of people and animals who make triggering noises.

**Foreign accent syndrome** is a speech impediment in which a person suddenly acquires a foreign accent after head trauma, stroke or migraine. At times speech returns to normal over time, with or without speech therapy.

**Fish odor syndrome (trimethylaminuria)** results from a genetic absence of a liver enzyme that normally clears trimethylamine, which smells like dead fish, from the blood. It is produced by bacterial fermentation in the colon. Since it's not degraded by the liver, when it builds up it makes the whole body smell. There is no cure.

**Fatal Familial Insomnia**, generally appearing between ages twenty and seventy, is a rare degenerative brain disorder, like the spongiform encephalopathy of mad cow disease, that results from an accumulation of a prion-related protein in the thalamus. The thalamus normally regulates unconscious bodily functions like breathing, sweating, heart rate, sleep and body temperature. Symptoms other than insomnia might include profuse sweating, pinpoint pupils, sudden menopause or impotence, dementia and muscle twitching. There is no cure.

**Fatal Constipation conditions:** (many think going a few days without a bowel movement is miserable, but such transient constipation is not fatal).

**Currarino syndrome** is a familial, at times fatal, condition causing constipation unresponsive to standard remedies, chronic bloating, recurrent fever and vomiting, usually since birth. A similar disorder, **Hirschsprung disease**, is a genetic absence, in parts of the colon, of nerve cells important for the colon's contractions. The absence of appropriate colonic muscular contraction and relaxation causes symptoms of constipation, abdominal swelling, vomiting, pain from

inflammation and failure to thrive in infants. It rarely starts in adulthood. Treatment consists of surgical removal of the abnormal colon segments.

congenital disorder and are corrected by supplementing those nutrients.

**Auto-Brewery Syndrome** occurs in rare people who, while eating diets high in sugar or carbohydrate, have overgrowths of certain colonic, oral or urinary organisms which ferment carbohydrate-rich foods into alcohol in sufficient quantities to cause intoxication, but insufficient to fail a drunk driving test. Our livers usually metabolize alcohol, so anyone with this disorder who also has liver disease would likely accumulate measurable blood alcohol levels. Other possible symptoms include belching, vomiting, irritable bowel syndrome, disorientation, fatigue and dizziness. Treatment includes antibiotics which are active against specific cultured organisms and probiotics to normalize the gut microbiome.

**LC-FAOD (Long chain fatty acid oxidation disorders)** are a group of rare, life-threatening genetic disorders caused by genetic defects of enzymes involved in delivering fatty acids to energy-producing organelles in cells, the mitochondria, to be 'burned' for energy. Because fatty acids are crucial energy sources throughout the body, symptoms may involve any organ, including the brain, liver, muscle, gastrointestinal system and heart. Times during which the person is unable to consume sufficient sugar and starchy food to compensate for the inability to use fat for energy typically trigger symptoms. Symptoms usually occur within hours of birth but may not appear until adulthood.

**Homocystinuria** is a rare inherited deficiency of the enzyme that normally turns the amino acid homocysteine into methionine. This causes a build-up of homocysteine in the blood, which is associated with premature coronary artery disease. Those with the congenital disorder are often tall, with a thin body with very high foot arches, seizures, intellectual disability, depression, early cataracts, various skeletal deformities, and retinal detachment or glaucoma. Late onset elevated homocysteine blood levels may also result from deficiencies of the vitamins folate, B2, B6 or B12, which don't cause the severe symptoms of the