

“It Stacks on Our Shoulders like Bricks”

Burden of Dystonia is More than a Movement Disorder

Dystonia is a neurological disorder that affects the physical body, but the impact goes far deeper. The hallmark signs of dystonia are excessive, involuntary muscle contractions that cause abnormal postures and/or repetitive movements. Individuals diagnosed with dystonia also commonly experience symptoms that affect more than how the body moves.

“Besides the physical motor symptoms of dystonia, many patients suffer from psychological complaints, such as anxiety and depression, but also problems with cognition, pain, and sleep have been reported. It appears that the type of dystonia predicts which non-motor symptoms are most common. However, there is a lot of overlap,” Professor Marina AJ de Koning-Tijssen is a movement disorders specialist in the Department of Neurology, University Medical Centre Groningen (Netherlands) and DMRF research grant recipient. Much of her work is devoted to better understanding non-motor features associated with dystonia. She explains: “Research showed that non-motor symptoms are an important burden for many patients with dystonia. Some studies showed that the non-motor symptoms even had a bigger influence on quality of life than the dystonic symptoms.”

Movement disorder neurologists are increasingly recommending that addressing these non-motor aspects of dystonia is essential for helping patients feel and function as well as possible. A team of specialists may be needed to implement a complete treatment plan.

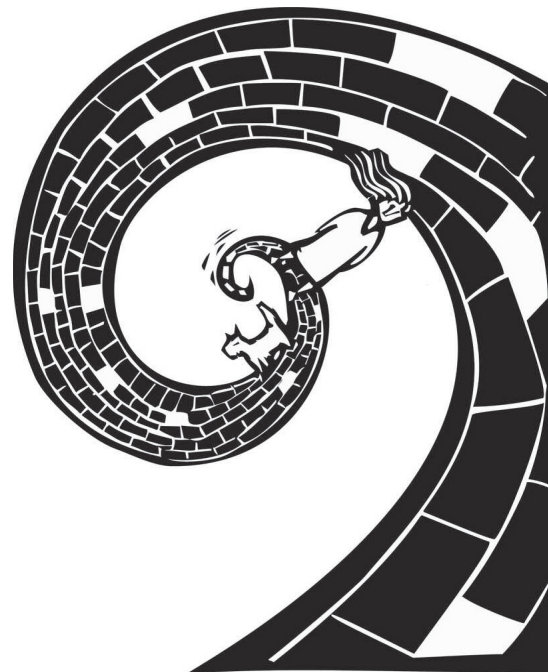
MENTAL HEALTH

The human brain is staggeringly complex: structures and pathways associated with movement are also involved in cognition, emotion, memory, and other mental functions. Research has demonstrated that individuals with dystonia are prone to certain mental health disorders.

“Whether these symptoms are part of the dystonia syndrome or are a consequence of living with dystonia is not totally clarified yet,” elaborates Dr. de Koning-Tijssen. “Nevertheless, researchers have found some clues that it is more likely that the non-motor symptoms are part of the dystonia syndrome.”

○ Depression & Anxiety

Individuals with dystonia are more likely to experience anxiety and depression than the general population, and at higher rates than individuals with other chronic disorders. Studies have shown that as many as 70% of focal and generalized dystonia patients will experience depression and/or anxiety over their lifetime. For individuals with cervical dystonia, this number may be as high as 90%. By comparison, 35% of people



in the general population will experience depressive and/or anxiety disorders at some point in their lives.

Patterns of psychiatric manifestations may vary depending on the type of dystonia. Individuals with cervical dystonia or dopa-responsive dystonia appear prone to both major depressive disorder and anxiety disorders. Anxiety disorders are especially prevalent in dystonia-affected musicians. Rates of depression are high among individuals with blepharospasm. By contrast, there may be little or no increased risk for depression and anxiety associated with spasmodic dysphonia/laryngeal dystonia compared to the general population.

It would seem intuitive that individuals with the most severe dystonia symptoms are more susceptible to depression

***“Having dystonia, or any disability or chronic condition, isn’t just about the symptoms. People tend to forget they are a whole person with anger and frustrations, guilt, depression, and it builds up. We aren’t just a product of a sudden condition.*”**

***“Therapy helped me learn to process what I was feeling both mentally and physically—it made me pay attention to what I was hanging on to, what was good around me, what to not waste my energy on, what to finally let go. I had to sort out the person I was before dystonia, and now with it. And it’s helped.*”**

“Our emotional state can affect our symptoms. We clench our necks, tighten our scalps when we cry, we curl our bodies inward. It stacks on our shoulders like bricks. We carry everything with us.”

*~Michaela Whitney,
DMRF Member*

and anxiety. In a study of mental health disorders among individuals with spasmodic dysphonia, severe voice impairment did seem to predict higher rates of depression and anxiety. Similarly, pain is highly correlated with depression in dystonia. But ironically, multiple studies have shown that the severity of dystonia is not a dominant influence on mental health. “In some types of dystonia the psychological complaints have no association with the severity of the dystonic motor symptoms,” says Dr. de Koning-Tijssen. “Therefore, it is unlikely that the psychological symptoms are a consequence of living with the difficulties of dystonia.”

Regardless of the severity of dystonia symptoms, the presence of depression and/or anxiety is among the most significant predictors of diminished quality of life. Because untreated mental health disorders can have serious and lasting health consequences, several research groups have recommended more routine evaluation of individuals diagnosed with dystonia for co-existing mood and anxiety disorders.

○ **Social Anxiety**

Social anxiety is the most common anxiety disorder among individuals with dystonia. One study showed that 50% of patients with cervical dystonia experience social anxiety, seemingly rooted in low self-esteem due to negative body image and not necessarily correlated to severity of dystonia symptoms.

○ **Alcoholism**

Myoclonus-dystonia is a risk factor for alcoholism, and individuals with cervical dystonia and isolated (primary) generalized dystonias may also be prone. Alcohol abuse in myoclonus-dystonia may be linked to obsessive-compulsive

disorder, a non-motor feature that appears uniquely common among individuals with this particular diagnosis but not dystonia more generally.

○ **Cognition**

“Problems with cognition are usually mild and have been reported in patients with inherited young onset dystonia and myoclonus-dystonia,” says Dr. de Koning-Tijssen. The source of cognitive changes can be challenging to assess, and research continues to explore this. Medications used to treat dystonia can cause cognition problems, especially affecting memory. Mood disorders can cause changes in executive functioning and ability to self-regulate. Disabling dystonia symptoms may make it difficult to concentrate, leading to attention deficits.

Several lines of evidence suggest mental health disorders are part of the underlying neurology of dystonia, not secondary to the dystonia or simply a coincidence. For example:

- Depression and anxiety often occur prior to the onset of dystonia.
- There may be no correlation between the severity of a person’s dystonia and psychiatric symptoms.
- Increased depression is seen in both asymptomatic DYT1 gene carriers and those with dystonia symptoms.
- Reduction in dystonia severity often does not alleviate depression or anxiety.

Furthermore, neurotransmitters associated with dystonia also have roles in psychological processes:

Continued on page 16

“Mention all symptoms connected to the dystonia to your neurologist, but especially depressive symptoms and anxiety are common—and important to address. Also sleep problems, fatigue, problems with alcohol consumption, and obsessions can be related to the dystonia.”

~ Professor Marina AJ de Koning-Tijssen, Department of Neurology, University Medical Centre Groningen

“Some researchers suggest that the non-motor symptoms result from an altered metabolism of signaling substances in specific regions in the brain. Two of those substances are dopamine and serotonin. Especially serotonin is known to play a role in psychological complaints but also in sleep.” Dr. de Koning-Tijssen recently earned a research contract through the DMRF’s Myoclonus-Dystonia Research Program to examine the role of serotonin in dystonia. Elze Timmers, PhD is integrally involved in the project.

Researchers acknowledge the complex interplay between a person’s movements, behaviors, and emotions. Dystonia is a formative experience; the impact can be life-changing. People naturally have an individual emotional and mental reaction to their circumstances. In some cases, it does appear that controlling dystonia motor symptoms has a positive effect on mental health. For example, a study in volunteers with cervical dystonia revealed decreased severity of depression with successful botulinum neurotoxin therapy.

“The main role for the neurologist is to recognize the non-motor symptoms. Then, adequate treatment can be started,” explains Dr. de Koning-Tijssen. “Treatment may be initiated by a neurologist, but most often this will be done by mental healthcare professionals with specific expertise in these areas.”

SLEEP

Problems with sleep are among the most common and problematic non-motor symptoms associated with dystonia. Although there is little evidence that dystonia causes daytime drowsiness, several studies do suggest dystonia interferes with the ability to sleep well. Fatigue is common and can be debilitating. Sleep disturbances occur even in cases when the dystonia symptoms are reduced or absent during sleep, and in cases where the dystonia is well-controlled with treatment.

In a study of blepharospasm and oromandibular dystonia patients, the more severe the dystonia, the greater the sleep disturbance. Interestingly, in a study of focal and generalized dystonia patients, trouble sleeping did not appear to correlate to severity of motor symptoms.

Abnormal brain plasticity during sleep may be implicated in the development of movement disorders, particularly dystonia and Parkinson’s disease. Plasticity is the brain’s ability to adapt and change over time, and sleep may have a role in reshaping brain processes involved in memory and learning. Dystonia has been linked to abnormal plasticity: learned movements that were once second nature—blinking, writing, walking—become abnormal. Researchers are beginning to explore the role of sleep in the brain’s learning processes, and how this may relate to the development of movement disorders.

PAIN

Not everyone with dystonia experiences pain, but depending on the type of dystonia, pain can be a pervasive and disabling symptom. Up to 76% of cervical dystonia patients have pain in the head, neck, and sometimes arm.

Pain can often be attributed to the repeated dystonic movements and abnormal postures. However, not all patients with similar symptoms experience the same degree of pain. This suggests the relationship between dystonia and pain may be more nuanced.

In many cases, alleviating the dystonia symptoms will reduce pain. For example, the sustained intense neck muscle contractions of cervical dystonia often cause muscle pain in the neck and shoulders as well as headache. The headache develops or worsens in relation to the cervical dystonia, and the location of the headache corresponds to the location of dystonic muscles. Alleviating the dystonic spasms can reduce both the muscle pain and secondary headache. However, people may experience pain that is more difficult to directly attribute to dystonia symptoms. Individuals with dystonic head tremor—a “no-no” or “yes-yes” shaking of the head—appear especially prone to headache, even if the tremor is mild, and the location of the headache is not clearly related to the muscle movement.

Researchers continue to explore the relationship between dystonia and pain. There may be neurological changes in how the body perceives and processes pain. Some research suggests that individuals with dystonia may have an altered threshold for pain, which has been measured even in body parts not affected by dystonia. Depression and sleep disturbance—which are common among dystonia patients—may worsen pain. Dystonia can also cause or worsen painful orthopedic conditions such as arthritis.

SENSORY FEATURES

The sensory nervous system is responsible for processing stimuli from the senses: sight, hearing, touch, taste, smell, and body awareness. “Research showed that the sensory function in the brain is altered in dystonia patients. However, the precise mechanism is not fully understood,” explains Dr. de Koning-Tijssen. Some of these sensory differences can be measured in the brain but not necessarily recognized by patients at a conscious level. For example, brain studies using neurophysiological techniques have shown that dystonia patients have trouble distinguishing certain types of visual and touch stimuli. However, some people do report sensory symptoms. For example, individuals with blepharospasm may experience light sensitivity. Sensory tricks, which are intentional movements or gestures that can temporarily reduce dystonia symptoms, are common and validate theories that dystonia is a disorder of how the nervous system integrates motor and sensory processes.

TREATING NON-MOTOR SYMPTOMS

Living well with dystonia requires more than addressing the physical movement symptoms. A team of healthcare professionals from multiple disciplines may be necessary, including a movement disorder neurologist, psychiatrist, clinical psychologist, and others depending on the needs of the individual. Appropriate complementary therapies may provide valuable underlying support to the treatment process.

In addition to the importance of individuals and families being mindful of non-motor symptoms associated with dystonia, “it is important that neurologists are aware of the existence of these non-motor symptoms,” says Dr. de Koning-Tijssen. “By doing additional research in order to understand more about the non-motor symptoms we hope to gain more attention for this important subject.”

Professor Marina AJ de Koning-Tijssen is head of the Movement Disorders section in the Department of Neurology, University Medical Centre Groningen, in the Netherlands. As a neurologist, she is an expert in hyperkinetic movement disorders including dystonia, myoclonus, hyperkplexia, tremor, tics, and functional jerks. After her Neurology training at Leiden University, with periods at the Johns Hopkins University in Baltimore and the Institute of Neurology and Neurosurgery, Queens Square, London, she started an internationally renowned movement disorders group, first in Amsterdam, and since 2012 at the University of Groningen. In 2016 the Groningen Expertise Centre for Rare Movements was established, officially part of the European Reference Network for Rare Neurological Diseases.

Anxiety

An anxiety disorder differs from simple worry in the following ways:

- It is more intense
- It is long-lasting
- It interferes with work, activities, or relationships

Social anxiety disorder is the fear of any situation where public scrutiny may occur, usually with the fear of embarrassment or humiliation.

Depression

Clinical depression includes at least one of these two symptoms, nearly every day, for more than two weeks:

- An unusually sad mood
- Loss of enjoyment and interest in activities that were once enjoyable

Additional symptoms of clinical depression can include:

- Lack of energy and tiredness
- Feeling worthless or feeling guilty
- Thinking about death or wishing to be dead
- Difficulty concentrating or making decisions
- Moving more slowly, or becoming agitated and unable to settle
- Sleeping difficulties, or sleeping too much
- Loss of interest in food, or eating too much; weight loss or weight gain

If you believe you may be experiencing symptoms of a mental health disorder, talk to your doctor about evaluation and treatment options.