Meet OMS Warrior ALEXA

Alexa

Alexa's battle began in the fall of 2009 at the age of 15 months. She toddled to bed one evening and did not walk again for months. During the early stages of illness, Alexa lost her ability to walk, sit up, talk, and even hold objects. Her parents took her to various doctors and specialists, but no one had a definitive answer for the rapid deterioration of Alexa’s health. Despite continuous efforts, her family was unable to find any answers. Finally, at the age of 4 years old, Alexa’s battle brought her family to a friend in Cincinnati who happened to be a doctor. He directed them to Texas Children’s Hospital in Houston, where, after a 3 months stay in the hospital, Alexa received her diagnosis of OMS.

Over the next 4 years, Alexa fought the battle of OMS. She endured countless scans, tests, and medical treatments. She also received speech, physical, and occupational therapies as she had to re-learn how to walk, talk, sit, grasp objects, and other developmental milestones. Alexa encountered numerous setbacks and struggles. She overcame them through her determination and perseverance. Despite her challenges at an early age, her family decided they needed to help others battling OMS. They founded the OMSLife Foundation in 2012 to provide information and be a resource; the foundation now serves more than 1,000 patients throughout the world. Our mission statement is “to raise awareness of Opsoclonus-Myoclonus Syndrome, maintain a support network for patients and caregivers, and fund research.”

Thank you to the OMS specialists who treated our OMS warriors, and a special thank you to NORD and Trio Health for their support.

General Discussion

Opsoclonus-myoclonus syndrome (OMS) is an inflammatory neurological disorder, often occurring as a paraneoplastic syndrome with neurological symptoms being the first sign of an occult tumor. OMS is characterized by associated ocular, motor, behavioral, sleep, and language disturbances. The onset is oftentimes abrupt and can be relatively severe, with the potential to become chronic unless the appropriate diagnosis and treatment are reached in a timely manner.

Signs and Symptoms

The component features of OMS include the presence of rapid, seemingly random eye movements in the horizontal, vertical, and diagonal directions (opsoclonus), an unsteady gait or inability to walk or stand (ataxia), and brief, repetitive, shiver-like muscle spasms or tremors within the arms, legs, or hands interfering with normal use (myoclonus). Behavioral and sleep disturbances, including extreme irritability, inconstantable crying, reduced or fragmented sleep (insomnia), and rage attacks are common. Difficulty articulating speech (dysarthria), sometimes with complete loss of speech and language, may occur. Additional symptoms, such as decreased muscle tone (hypotonia) and vomiting, have also been noted.

Causes

The most common cause of OMS in young children is an occult (ie, small, often hidden) tumor. The symptoms of OMS, as a paraneoplastic syndrome, presumably stem from the immune system attacking the tumor, leading to secondary inflammatory effects on the central nervous system. Despite the primarily neurological status of this condition, tumors are generally not found in the brain, but rather in other areas of the body, such as the chest or abdomen. In 50% to 80% of affected young children, a tumor of the immature nerve cells (neuroblastoma or ganglioneuroblastoma) is responsible for the development of symptoms associated with OMS. There is, however, also a relatively high rate of spontaneous tumor regression in this population, meaning the tumor may be gone by the time OMS is diagnosed and investigations are done to look for it. In contrast, among older children or teens, the disorder is often attributed to various, predominantly viral infections. When OMS occurs in adults, it is often as a paraneoplastic syndrome related to other kinds of tumors; most cases are due to lung cancers or breast cancers. Other unusual, often hidden (paraneoplastic) tumors are responsible in a few cases. In 50% to 80% of affected young children, a tumor of the immature nerve cells (neuroblastoma or ganglioneuroblastoma) is responsible for the development of symptoms associated with OMS. It is because of Alexa’s challenges at an early age that her family decided they needed to help others battling OMS. They founded the OMSLife Foundation in 2012 to provide information and be a resource; the foundation now serves more than 1,000 patients throughout the world. Our mission statement is “to raise awareness of Opsoclonus-Myoclonus Syndrome, maintain a support network for patients and caregivers, and fund research.”

continued on page 15
Audrey's battle with OMS began in February 2010 when she was 7. She had just fought back-to-back bouts of strep throat when one morning her parents noticed her hand was shaking slightly. The next day she started waking up in the middle of the night in a screaming rage. She had a very hard time keeping her balance, and the rage fits became a daily event and her ability to focus was lost. Audrey's parents continued to look for a solution until they found Dr. Mark Gorman at Boston Children's Hospital. He was one of the few specialists in the United States who treated OMS, and when they met with him, he immediately told them that Audrey suffered from OMS. Although her parents were relieved to finally have a correct diagnosis, the treatment was non-existent, as there was none for OMS.

Audrey had tried regular IVIg treatments, speech and physical therapy, and took numerous medications to help her function with her condition. However, her condition did not improve and her parents were getting frustrated as their household was in complete disarray. They did not know how to handle the rage attacks and Audrey's loss of balance; to make matters worse, she also developed severe anxiety and OCD.

Audrey's parents continued to look for a solution until they found Dr. Mark Gorman at Boston Children's Hospital. He was one of the few specialists in the United States who treated OMS, and when they met with him, he immediately told them that Audrey suffered from OMS. Although her parents were relieved to finally have a correct diagnosis, the treatment was non-existent, as there was none for OMS.

About 95% of children with OMS are initially diagnosed as having acute cerebellar ataxia, a much more common disorder characterized by sudden, uncoordinated movements originating from disease or injury to the cerebellum. In children with OMS, the ataxia may appear before the eye findings, leading to this particular misdiagnosis. Where present, the existence of opsoclonus is consistent with acute cerebellar ataxia, and OMS should be suspected as the true diagnosis. It should be noted that occurrences of opsoclonus or myoclonus may not be constant and can be subtle, making these findings harder to recognize and properly diagnose.

Myoclonus and tremor may also occur in other conditions without opsoclonus, both with or without ataxia. Myoclonus, in particular, may accompany a number of neurologic diseases, including seizure disorders, brain injuries, hereditary brain diseases, viral infections, metabolic disorders, and toxicological illness. (More information on these disorders can be found by searching for “general myoclonus” in the Rare Disease Database.)

OMS is a rare disorder. It affects 1 per million individuals worldwide, usually infants and young children, although it has also been known to affect adults. The peak age of onset in children is about 18 months of age, with very few cases diagnosed before 12 months. In rare instances, OMS can also occur in somewhat older children, up to approximately 6 years of age. Occurrence of opsoclonus in infants under 6 months old is quite uncommon, and opsoclonus in that age group, when isolated, is usually found to originate from another cause. OMS is thought to be slightly more common in girls than boys. While 50% of children with OMS are found to have a neuroblastoma or similar tumor, only 3% of children with such tumors present with OMS, though a small percentage more can go on to develop the condition later in their course of treatment.
Braeden OMS became obvious in March 2007. At just shy of 2 years old, he went to bed one night seemingly fine. The next morning he was unable to stand without falling and unable to walk without stumbling or falling into things. The changes were radical and terrifying. Braeden’s mom and dad had been taught to recognize the symptoms of OMS and knew just what their son was seeing. They immediately called their OMS specialist and began the process of getting Braeden into the OMS specialist’s office and scheduled for a lumbar puncture to make the diagnosis of OMS.

Braeden’s parents were told he had OMS and needed to be treated with high-dose corticosteroids for a long period of time to help develop individual educational accommodations. His doctors did not know what would happen to him and his family in the future. He had to take into account the other illness he also had to treat for OMS and another to treat the cancer that caused the OMS.

Diagnosis

The diagnosis of OMS is clinical; there is no single diagnostic test that serves as a specific marker of the disease, as the exact immunological factors responsible for causing the syndrome remain unidentified. The combined presence of the “dancing eyes,” the shock-like muscle spasms, and the gait impairment, especially if accompanied by irritability, are highly reliable indicators of this syndrome. To detect a tumor in children, either a CT scan or an MRI with contrast of the chest, abdomen, and pelvis should be done. An MRI with contrast of the brain should be performed in all patients. MIBG or PET scanning should also be done in all patients with OMS as an important second investigation looking for occult tumors. In addition, a spinal tap to detect neuroinflammation is necessary. Besides routine tests for infection, recommended cerebrospinal fluid (CSF) studies include oligoclonal bands (with paired serum sample), to see if there are antibodies secreted by B-cells in the CSF. If available at the institution, CSF lymphocyte subset analysis (flow cytometry) using immunophenotyping may identify increased frequency of CSF CD 19+ B-cells, an important biomarker of OMS disease activity. Various nonspecific autoantibodies in some children with OMS have also been detected in research laboratories.

Standard Therapies

TREATMENT

Early and aggressive treatment for OMS is recommended, as some evidence suggests this better achieves the goal of a durable and more complete neurological remission. If a tumor is present, surgical resection is standard. The tumors in young children are usually low-stage neuroblastomas or ganglioneuroblastomas (stage I or II), and chemotherapy or radiation therapy is generally not indicated. Tumor resection does not typically provide sufficient clinical benefit for OMS.

OMS treatment, which is usually continued for at least 1 to 2 years, should involve combined immunotherapies. While evidence is limited, the expert consensus for treatment involves a 3-agent protocol, with initial use of intravenous immunoglobulin (IVIg), rituximab, and either high-dose corticosteroids (IV methylprednisolone followed by pulse oral dexamethasone) or ACTH (corticotropin) appearing to have the best-documented outcomes for moderate and severe cases. Most children respond initially to rituximab, and either high-dose corticosteroids or ACTH, showing at least partial improvement. Those treated with steroids or ACTH alone, how-

ever, tend to promptly relapse when treatment is stopped. Over time, it is worth noting that treatments with corticosteroids or ACTH may have substantial cortisol-related adverse effects that should be monitored carefully, including weight gain, hypertension, and reduction in bone density. For OMS relapse, low-dose IV cyclophosphamide (3 to 6 cycles) or repeated courses of rituximab (1 to 2 cycles) may be given. Oral weekly methotrexate may be a useful steroid-sparing agent in chronic relapsing-remitting disease. Along with the above noted pharmacological agents, all children should receive appropriate physical, occupational, and speech therapy services. Formal neurodevelopmental and neuropsychological assessments should be performed over time to help develop individual educational accommodations.
Chloe, born a healthy baby girl in 1992, developed normally and met the child development milestones until the age of 18 months. After a flu and febrile episode following her 18-month vaccination against MMR, however, Chloe started to show signs of ataxia. Her parents took her to the doctor, who ordered an MRI of Chloe’s head. The MRI results were concerning, and the doctors wanted to rule out any other diagnoses, including OMS.

Chloe’s symptoms worsened, and tests continued. Eventually, she was diagnosed with OMS. To help Chloe regain her lost coordination, she had speech therapy from 3 years old until 6 years old, and physical and occupational therapy from 1 year old until 11 years old. She also took karate, ballet, and horseback riding lessons. As a result of the OMS attacking Chloe’s brain, she met with learning disabilities. Though she participated in special education programs in school, Chloe was mainstreamed into regular classrooms throughout her school career. Her Individual Education Plan (IEP) was essential to receive the accommodations to which she was entitled; these included audio books, extended testing times, additional time on assignments, and modified assignments because of two parents advocating for her. Through her teachers and meeting with her teachers regularly, Chloe was able to excel during her academic career. She graduated with honors from college with an Associate Degree in Early Childhood Education and is currently a full-time teacher and living independently.

Investigational Therapies

Information on current clinical trials is posted on the Internet at www.clinicaltrials.gov. All studies receiving US government funding, and some supported by private industry, are posted on this government web site.

For information about clinical trials being conducted at the NIH Clinical Center in Bethesda, MD:
Toll free: (800) 411-1222
TTY: (866) 411-1010
Email: prpl@cc.nih.gov

Current clinical trials are also posted on the NORD website:
www.rarediseases.org

For information about clinical trials sponsored by private sources, contact:
www.centerwatch.com

For information about clinical trials conducted in Europe, contact:
www.clinicaltrialsregister.eu

What is OMS?

continued from page 17

OUTCOME

Almost all children with neuroblastomas and OMS survive their tumor, which usually does not behave aggressively, though larger tumors may pose difficulties for resection. In contrast, the tumors associated with OMS in adults are often aggressive and can be fatal. The OMS relapse rate in children treated with only conventional agents is 50% to 75%. Increased immunosuppression has, however, improved neurodevelopmental outcomes in OMS. With more aggressive initial therapies in children, the relapse rate appears to be much lower. OMS onset in the first 2 years of life is particularly damaging to expressive speech and language development, and may result in a higher incidence of residual cognitive impairment. The best outcomes appear to be those involving early combination therapy and mild-to-moderate OMS severity. Failure to achieve complete neurological remission and multiple relapses may result in chronic-progressive OMS, with permanent deficits, such as attention deficit disorder (ADD), attention-deficit/hyperactivity disorder (ADHD), obsessive-compulsive disorder (OCD), and irreversible cognitive impairment (low IQ). Children in the chronic state may become oppositional, depressed, or aggressive, and attention to these issues can help improve quality of life (QoL). Parents with a severely ill infant or child may develop “fragile child syndrome” and have difficulty ever seeing their child as a normal, thriving individual, with “ordinary” behavioral issues of childhood misinterpreted to represent a relapse of OMS. These parents may benefit from counseling to gradually adjust the management of their child’s ongoing behavioral and developmental challenges.
Meet OMS Warrior COLE

COLE

Cole

Cole was diagnosed with a malignanc and OMS in July 2011, when he was 15 months old. His symptoms started with a viral infection and fever, and he went from a happy, playful baby to an irritable toddler who could not sleep and could not walk. After the first day, he also started having hand tremors, body tremors, and eye movement problems. He became really tired and had difficulty walking. It was heartbreaking to watch. He had even quit babbling in the mornings and rising to greet us in his crib.

Our world came crashing down as we were given his diagnosis. We were told that most children with OMS have developmental delays and behavioral issues. Our son and family would be facing a long and challenging road with long-term deficits and relapses. We were also told that OMS is a rare condition that did not have much funding for research and for well-studied treatment protocols. With the support of the staff and doctors at Dell Children’s and the courage and support from many OMS Warriors around the world, we are able to deal with the challenges of OMS.

After several years of speech, occupational, and physical therapies, together with periods of isolation due to being immunocompromised, Cole is teaching us an amazing lesson. He is a public school in a regular class with accommodations and is at or above grade level in many classes. He is loved by all who know him and has many friends. Cole is involved in many activities, such as Cub Scouts, golf, and basketball. He dreams of one day going to his father’s alma mater, University of Kentucky, and playing basketball. We still hold our breath if we hear him cough or notice him tripping or that he is too quiet, but we have never lost hope. With every sad tear we have shed, we have also shed tears of happiness. Happiness for watching our son do some of the simplest things.

Cole is involved in many activities, such as Cub Scouts, golf, and basketball. He dreams of one day going to his father’s alma mater, University of Kentucky, and playing basketball. We still hold our breath if we hear him cough or notice him tripping or that he is too quiet, but we have never lost hope. With every sad tear we have shed, we have also shed tears of happiness. Happiness for watching our son do some of the simplest things.

We have had Cole and his family as part of our 2012 Sarcoma Awareness Month. We are very grateful for their honesty and strength and their willingness to share their story with us. We hope that other parents and caregivers of children with OMS and other rare conditions can find inspiration and support from Cole and his family.

For more information about the OMSLife Foundation, please go to our website at www.omslifefoundation.org or follow us on Facebook at www.facebook.com/OMSLifeInformation.
Meet OMS Warrior LAUREN

Lauren

In January 2009, we were developing 21-month-old daughter began having difficulty balancing and within a couple days had stopped smiling to kiss her, as if to oneself. She would go to sleep and wake up as if driving a car, unable to do due to tremors and ataxia. She had her audibility function at the time of diagnosis. She had difficulty making any fine motor movements. She was seen by Dr. Mark Gorman at Boston Children’s Hospital. A brain MRI was performed and showed neuroaxial films of the cerebellum, which noted mild ataxia. 

Lauren showed mild improvements and began physical and occupational therapies. After she relapsed in April 2009 and May 2009, we were told she had elevated HVA/VMA catecholamine levels, possibly indicating neuroblastoma. Six months after her symptoms initiated, Dr. Mark Gorman diagnosed her with OMS.

April 2012. As of 2013, she had fully recovered from the complications and surgery. In March 2012 she underwent an 8-hour surgery to hopefully bypass the complications she was having and spent 11 days in the PICU in a ventilator, due to further complications. Lauren finally returned home in June 2012.

Lauren continued her battle with OMS with the help of CellCept® and CellCept®. The dosage has been slowly increased since November 2011 and has been slowly increased to 100 mg per day. Currently, she is taking 75 mg a day and was born off CellCept®. Lauren is on her second port and her veins have not recovered from the excessive use. Since she was first diagnosed.

Despite her OMS, Lauren is a well-adjusted child who remains engaging, friendly, and kind. She is in a regular education environment but was held back one year. She is on grade level for academics and social and emotional development and mostly struggles with fatigue and her effects after every IVIG treatment.

Leading specialists in treating OMS across the United States agree this therapy produces the best results known at this time. Use of only one or two of these options, or administering all of them at sub-therapeutic doses, will typically deliver sub-optimal results.

The treatment protocol should be accompanied by nonpharmaceutical modalities, including

- Speech therapy
- Physical therapy
- Behavioral therapy
- Occupational therapy

Long-term cognitive and physical issues are likely with delayed treatments and therapies. It is therefore highly recommended that physicians who are inexperienced in treating OMS coordinate the protocol with known specialists. Severity of the disease and time for proper treatment are directly correlated with long-term QoL for the patient.

Our call-to-action is straightforward:

CLINICIANS – Align your efforts with a specialist in OMS to ensure best results. Be aggressive with treatments and therapies, and if in doubt, direct the patient to the specialist.

RESEARCHERS – Align efforts with the International OMS Steering Committee, and collaborate with the committee to coordinate bio-samples.

CAREGIVERS AND PATIENTS – Get educated about OMS in order to understand the disease and its treatment protocols. Consider a second opinion if the prescribed protocol is outside of typical trends by those specializing in OMS. Pay your knowledge forward by participating in the NORD/OMSLife Patient Registry as well as in other patient clinical registries and bio-banks.

The treatment protocol should be accompanied by nonpharmaceutical modalities, including

- Speech therapy
- Physical therapy
- Behavioral therapy
- Occupational therapy

Long-term cognitive and physical issues are likely with delayed treatments and therapies. It is therefore highly recommended that physicians who are inexperienced in treating OMS coordinate the protocol with known specialists. Severity of the disease and time for proper treatment are directly correlated with long-term QoL for the patient.

Our call-to-action is straightforward:

CLINICIANS – Align your efforts with a specialist in OMS to ensure best results. Be aggressive with treatments and therapies, and if in doubt, direct the patient to the specialist.

RESEARCHERS – Align efforts with the International OMS Steering Committee, and collaborate with the committee to coordinate bio-samples.

CAREGIVERS AND PATIENTS – Get educated about OMS in order to understand the disease and its treatment protocols. Consider a second opinion if the prescribed protocol is outside of typical trends by those specializing in OMS. Pay your knowledge forward by participating in the NORD/OMSLife Patient Registry as well as in other patient clinical registries and bio-banks.
Michelle's journey with OMS began in 2010 when she was a 24-year-old graduate school student. Starting Slowly: a change in her vision prompted her to visit an eye doctor. After being diagnosed with OMS, Michelle continued her studies, and her symptoms slowly returned in the following months. Throughout the years, she experienced a variety of symptoms, including nausea/vomiting, and extremely painful and contorting muscle spasms. Despite months of ER visits and specialist visits, she did not receive a diagnosis. Michelle continued her education and received her doctorate degree and has maintained part-time work in the physiology research field. She also enjoys rock climbing and skiing with her family.

Michelle’s journey with OMS began in 2010 when she was a 24-year-old graduate school student. Starting Slowly: a change in her vision prompted her to visit an eye doctor. After being diagnosed with OMS, Michelle continued her studies, and her symptoms slowly returned in the following months. Throughout the years, she experienced a variety of symptoms, including nausea/vomiting, and extremely painful and contorting muscle spasms. Despite months of ER visits and specialist visits, she did not receive a diagnosis. Michelle continued her education and received her doctorate degree and has maintained part-time work in the physiology research field. She also enjoys rock climbing and skiing with her family.

When she traveled from her home in Oregon to New York, the neurologist diagnosed her with OMS and Michelle connected with an adult diagnosed with OMS who recommended she see the same provider. Tests showed high levels of white blood cells in her CSF, thus indicating an autoimmune component to her mysterious symptoms. Yet everything appeared normal. Specialists remained stumped and they would blame it on stress or say she was overthinking her symptoms. A referral to a local hospital led to a diagnosis of OMS, and her symptoms improved. During this time, she continued to connect with other adults and parents of OMS warriors. Though she supports families in similar situations, she found it challenging to find other adults with OMS to receive ongoing treatment due to provider and insurance resistance. Over the following 7 years, Michelle experienced ups and downs fighting insurance denials for IVIg and Rituximab. Moving their home required establishing new physician relationships, and the process of educating them and ruling out other conditions began yet again.

Michelle continued to receive IVIg treatment, although she had great improvements with the IVIg, her symptoms slowly returned in the following months. Thankfully, Michelle now lives a rewarding life as a mom with her family and dogs. She completed a neuroscience degree and has received a part-time position in the physiology research field. She also enjoys rock climbing and along with her family.

Meet OMS Warrior MICHELLE

Michelle

Michelle’s journey with OMS began in 2010 when she was a 24-year-old graduate school student. Starting Slowly: a change in her vision prompted her to visit an eye doctor. After being diagnosed with OMS, Michelle continued her studies, and her symptoms slowly returned in the following months. Throughout the years, she experienced a variety of symptoms, including nausea/vomiting, and extremely painful and contorting muscle spasms. Despite months of ER visits and specialist visits, she did not receive a diagnosis. Michelle continued her education and received her doctorate degree and has maintained part-time work in the physiology research field. She also enjoys rock climbing and skiing with her family.

When she traveled from her home in Oregon to New York, the neurologist diagnosed her with OMS and Michelle connected with an adult diagnosed with OMS who recommended she see the same provider. Tests showed high levels of white blood cells in her CSF, thus indicating an autoimmune component to her mysterious symptoms. Yet everything appeared normal. Specialists remained stumped and they would blame it on stress or say she was overthinking her symptoms. A referral to a local hospital led to a diagnosis of OMS, and her symptoms improved. During this time, she continued to connect with other adults and parents of OMS warriors. Though she supports families in similar situations, she found it challenging to find other adults with OMS to receive ongoing treatment due to provider and insurance resistance. Over the following 7 years, Michelle experienced ups and downs fighting insurance denials for IVIg and Rituximab. Moving their home required establishing new physician relationships, and the process of educating them and ruling out other conditions began yet again.

Michelle continued to receive IVIg treatment, although she had great improvements with the IVIg, her symptoms slowly returned in the following months. Thankfully, Michelle now lives a rewarding life as a mom with her family and dogs. She completed a neuroscience degree and has received a part-time position in the physiology research field. She also enjoys rock climbing and along with her family.

Meet OMS Warrior MICHELLE

Michelle’s journey with OMS began in 2010 when she was a 24-year-old graduate school student. Starting Slowly: a change in her vision prompted her to visit an eye doctor. After being diagnosed with OMS, Michelle continued her studies, and her symptoms slowly returned in the following months. Throughout the years, she experienced a variety of symptoms, including nausea/vomiting, and extremely painful and contorting muscle spasms. Despite months of ER visits and specialist visits, she did not receive a diagnosis. Michelle continued her education and received her doctorate degree and has maintained part-time work in the physiology research field. She also enjoys rock climbing and skiing with her family.

When she traveled from her home in Oregon to New York, the neurologist diagnosed her with OMS and Michelle connected with an adult diagnosed with OMS who recommended she see the same provider. Tests showed high levels of white blood cells in her CSF, thus indicating an autoimmune component to her mysterious symptoms. Yet everything appeared normal. Specialists remained stumped and they would blame it on stress or say she was overthinking her symptoms. A referral to a local hospital led to a diagnosis of OMS, and her symptoms improved. During this time, she continued to connect with other adults and parents of OMS warriors. Though she supports families in similar situations, she found it challenging to find other adults with OMS to receive ongoing treatment due to provider and insurance resistance. Over the following 7 years, Michelle experienced ups and downs fighting insurance denials for IVIg and Rituximab. Moving their home required establishing new physician relationships, and the process of educating them and ruling out other conditions began yet again.

Michelle continued to receive IVIg treatment, although she had great improvements with the IVIg, her symptoms slowly returned in the following months. Thankfully, Michelle now lives a rewarding life as a mom with her family and dogs. She completed a neuroscience degree and has received a part-time position in the physiology research field. She also enjoys rock climbing and along with her family.
Meet OMS Warrior ZARA

Zara

Zara’s life with OMS began in May 2014 when she underwent a sudden change in her behavior, both physically and mentally. She lost her ability to sit up and maintain her balance that she had been able to do since it was diagnosed with OMS. While waiting for her appointment at PCH, Zara continued to struggle to maintain the balance and she was constantly tired due to the fatigue. After a visit to Florida with her OMS specialist in July 2016 and after following her aggressive treatment measures, Zara improved drastically. Five weeks after that July visit she began fully active and now is 5 years old and has been through this journey.

At the OMS Caregivers Conference and effective as it should have been. We thought she was making progress, but in reality she was not. Due to the rareness of OMS and lack of experience treating it, Zara’s initial therapy was not as aggressive in her pelvis area, and she was diagnosed with OMS.

Zara’s life with OMS began in May 2014 when she underwent a sudden change in her behavior, both physically and mentally. She lost her ability to sit up and maintain her balance that she had been able to do since it was diagnosed with OMS. While waiting for her appointment at PCH, Zara continued to struggle to maintain the balance and she was constantly tired due to the fatigue. After a visit to Florida with her OMS specialist in July 2016 and after following her aggressive treatment measures, Zara improved drastically. Five weeks after that July visit she began fully active and now is 5 years old and has been through this journey.

OMS in the REAL WORLD

Early and accurate diagnosis is important for these young patients because of potential detrimental effects of disease on neurodevelopment. Medications for treatment of OMS include IVIg, corticosteroids/ACTH, chemotherapies, and immunotherapies, and clinical data suggest that combination therapy of corticosteroids/ACTH, IVIg, and rituximab may be useful in preventing neurodevelopmental damage. In addition to medications, non-pharmacological therapies are efficaciously prescribed to address speech, physical, occupational, and behavioral challenges. In many cases, treatment of OMS with the medications and therapies will typically take from 1 to 5 years to get the patient to baseline and in remission. During treatment or in the absence of disease control, medications to treat symptoms are commonly employed (e.g., sleep aids, proton pump inhibitors, mental health medications).

The outlook for patients with OMS is promising provided the continued trend of early diagnosis and early treatment. The main challenge to effective intervention is awareness, and the difficulty may be decreased through continued efforts of NORD, OMSLife, and publication of patient data from the OMS Natural History register.

Factors that might be contributing to these positive changes include greater awareness of the disease by medical personnel, greater collaboration mechanisms for congenital and pediatric neuro-musculo-skeletal, and child neuro-musculo-skeletal (OMS, BWS, and other syndromes). The outlook for patients with OMS is promising provided the continued trend of early diagnosis and early treatment. The main challenge to effective intervention is awareness, and the difficulty may be decreased through continued efforts of NORD, OMSLife, and publication of patient data from the OMS Natural History registry.

OMS in the REAL WORLD

Early and accurate diagnosis is important for these young patients because of potential detrimental effects of disease on neurodevelopment. Medications for treatment of OMS include IVIg, corticosteroids/ACTH, chemotherapies, and immunotherapies, and clinical data suggest that combination therapy of corticosteroids/ACTH, IVIg, and rituximab may be useful in preventing neurodevelopmental damage. In addition to medications, non-pharmacological therapies are efficaciously prescribed to address speech, physical, occupational, and behavioral challenges. In many cases, treatment of OMS with the medications and therapies will typically take from 1 to 5 years to get the patient to baseline and in remission. During treatment or in the absence of disease control, medications to treat symptoms are commonly employed (e.g., sleep aids, proton pump inhibitors, mental health medications).

The outlook for patients with OMS is promising provided the continued trend of early diagnosis and early treatment. The main challenge to effective intervention is awareness, and the difficulty may be decreased through continued efforts of NORD, OMSLife, and publication of patient data from the OMS Natural History register.

Factors that might be contributing to these positive changes include greater awareness of the disease by medical personnel, greater collaboration mechanisms for congenital and pediatric neuro-musculo-skeletal, and child neuro-musculo-skeletal (OMS, BWS, and other syndromes). The outlook for patients with OMS is promising provided the continued trend of early diagnosis and early treatment. The main challenge to effective intervention is awareness, and the difficulty may be decreased through continued efforts of NORD, OMSLife, and publication of patient data from the OMS Natural History registry.

Factors that might be contributing to these positive changes include greater awareness of the disease by medical personnel, greater collaboration mechanisms for congenital and pediatric neuro-musculo-skeletal, and child neuro-musculo-skeletal (OMS, BWS, and other syndromes). The outlook for patients with OMS is promising provided the continued trend of early diagnosis and early treatment. The main challenge to effective intervention is awareness, and the difficulty may be decreased through continued efforts of NORD, OMSLife, and publication of patient data from the OMS Natural History registry.

Factors that might be contributing to these positive changes include greater awareness of the disease by medical personnel, greater collaboration mechanisms for congenital and pediatric neuro-musculo-skeletal, and child neuro-musculo-skeletal (OMS, BWS, and other syndromes). The outlook for patients with OMS is promising provided the continued trend of early diagnosis and early treatment. The main challenge to effective intervention is awareness, and the difficulty may be decreased through continued efforts of NORD, OMSLife, and publication of patient data from the OMS Natural History registry.

Factors that might be contributing to these positive changes include greater awareness of the disease by medical personnel, greater collaboration mechanisms for congenital and pediatric neuro-musculo-skeletal, and child neuro-musculo-skeletal (OMS, BWS, and other syndromes). The outlook for patients with OMS is promising provided the continued trend of early diagnosis and early treatment. The main challenge to effective intervention is awareness, and the difficulty may be decreased through continued efforts of NORD, OMSLife, and publication of patient data from the OMS Natural History registry.

Factors that might be contributing to these positive changes include greater awareness of the disease by medical personnel, greater collaboration mechanisms for congenital and pediatric neuro-musculo-skeletal, and child neuro-musculo-skeletal (OMS, BWS, and other syndromes). The outlook for patients with OMS is promising provided the continued trend of early diagnosis and early treatment. The main challenge to effective intervention is awareness, and the difficulty may be decreased through continued efforts of NORD, OMSLife, and publication of patient data from the OMS Natural History registry.