MOWAT-WILSON SYNDROME FOUNDATION®

Supporting Rare and Special Families

WELCOME PACKET

The Mowat-Wilson Syndrome Foundation
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Greetings!

The Board of Directors of the Mowat-Wilson Syndrome Foundation would like to welcome you and your family to our MWS family!

We know you have a lot of questions and are not sure where to begin. The foundation is here to be a resource for you. Please visit our website at www.mowat-wilson.org where you will find the latest information on MWS, past and upcoming MWS events, and the MWS newsletter.

We invite each of you to sign up for the MWS newsletter, join in the various MWS Facebook pages, volunteer at any of the MWSF events, donate to the Foundation, and/or lead a MWS fundraiser.

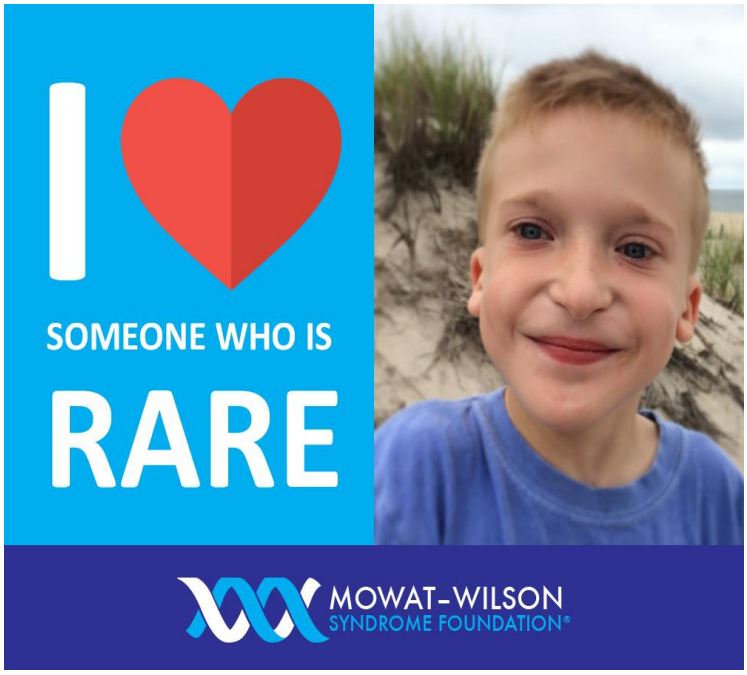
Included in this packet, you will find a letter to doctors that you can use when you see a pediatrician, general practitioner, specialist, and when you have emergency room or urgent care visits. We have also included a few other suggestions that have been helpful to board members along the way. These are suggestions that we as the board have found to be valuable to us after many years of learning to be caregivers to our MWS children and grandchildren.

The Board of Directors is made up of MWS family members and others who are truly invested in the mission of the Foundation and are all volunteers. The MWS Foundation also has a Medical Advisory Board made up of doctors and researchers in MWS and related fields, and a Community Advisory Board made up of MWS Caregivers.

Welcome and we hope to meet you at an event!

Sincerely,

The Mowat-Wilson Syndrome Foundation Board of Directors



Many children and adults with MWS need referrals to specialists including genetics, neurology, cardiology, gastroenterology, nephrology and various therapies including physical, occupational, and speech therapy.

Mowat-Wilson syndrome is often associated with:

- Microcephaly
- Seizures
- Structural brain abnormalities, such as Agenesis of the Corpus Callosum
- Intellectual disability ranging from moderate to severe
- Vision abnormalities, such as CVI and eye misalignment
- Absent or severely impaired speech. If speech develops, it is delayed until mid-childhood or later.
- Higher receptive language than expressive language
- Heart defects
- Delayed development of motor skills such as sitting, standing, and walking.
- Hirschsprung disease in greater than 50% of diagnosed cases, associated with severe constipation, intestinal blockage, and enlargement of the colon. Chronic constipation also occurs frequently in people with Mowat-Wilson syndrome who have not been diagnosed with Hirschsprung disease.
- Abnormalities of the urinary tract and genitalia
- Less commonly, this condition can also affect teeth, hands, and skin coloring (pigmentation)

www.mowat-wilson.org

Dear Practitioner,

This letter provides you with a summary of Mowat-Wilson Syndrome (MWS)

MWS is a genetic condition that affects many parts of the body. Major signs of this disorder frequently include distinctive facial features, intellectual disability, delayed development, an intestinal disorder called Hirschsprung disease, and other birth defects.

Children with MWS have a square-shaped face with deep-set, widely spaced eyes. They also have a broad nasal bridge with a rounded nasal tip; a prominent and pointed chin; large, flaring eyebrows; and uplifted earlobes with a dimple in the middle. These facial features become more distinctive with age, and adults with MWS have an elongated face with heavy eyebrows and a pronounced chin and jaw. Affected people tend to have a smiling, open- mouthed expression, and they typically have friendly and happy personalities.

Although many different medical issues have been associated with MWS, not every individual with this condition has all of these features. MWS is caused by mutations in the ZEB2 gene. This gene provides instructions for making a protein that plays a critical role in the formation of many organs and tissues before birth. MWS almost always results from a loss of one working copy of the ZEB2 gene in each cell.

In some cases, the entire gene is deleted. In other cases, mutations within the gene lead to the production of an abnormally short, nonfunctional version of the ZEB2 protein. A shortage of this protein disrupts the normal development of many organs and tissues, which causes the varied signs and symptoms of MWS.

A comprehensive study published in 2020 in the Orphanet Journal of Rare Diseases provides the first up-to-date growth charts specific for infants and children of different ancestries. Data was collected through collaborations with the Italian MWS association (AIMW) and the MWS Foundation. Height, weight, body mass index (BMI), and head circumference were compared to those from standard international growth charts for healthy children. These growth charts should assist pediatricians and caregivers in providing optimal care to MWS patients with problems related to physical growth.

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FAQS

What is Mowat-Wilson Syndrome (MWS)?

MWS is a genetic condition that affects many parts of the body, including distinctive facial features, intellectual disability, delayed development, an intestinal disorder called Hirschsprung disease, and other birth defects.

What causes MWS?

MWS is caused by a mutation in the ZEB2 gene. It typically occurs for the first time in a person with MWS and is not inherited from a parent. The mutation occurs during the formation of reproductive cells (eggs or sperm) or in early embryonic development.

How is MWS diagnosed?

Diagnosis for a genetic or rare disease can often be challenging and healthcare professionals typically look at medical history, symptoms, physical exam, and laboratory test results in order to make a diagnosis.

What is the prognosis of MWS?

There is little information about the long-term outlook for people with MWS. Quality of life and life expectancy depend on the presence and severity of birth defects.

What is the treatment for MWS?

Treatment for MWS depends on the symptoms present and focuses on the specific needs of each person.

How common is MWS?

According to Orphanet, prevalence is estimated at 1/50,000- 70,000 live births.

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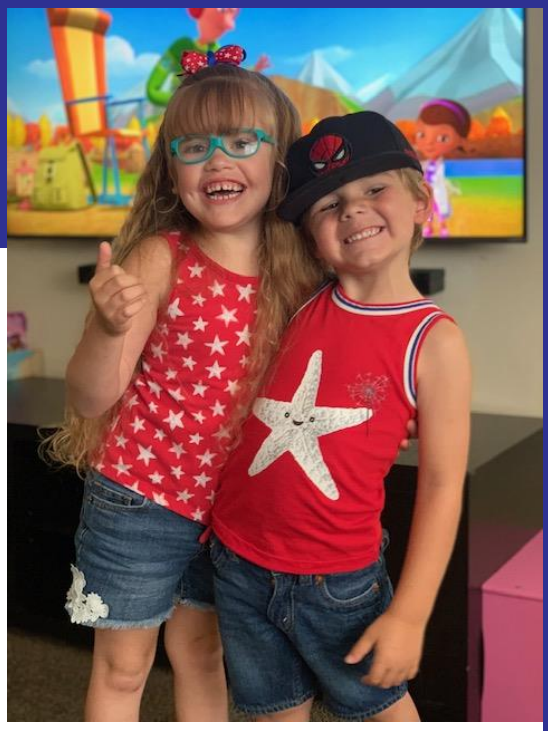
DEAR CAREGIVERS,



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Being a caregiver is often like being the "Lone Ranger." It may be difficult to keep up with or reach out to family and friends, and it may feel like these relationships are fading. Generally, caregivers are more likely to feel isolated if they cannot leave their home, have experienced a major life change (such as the loss of a job), live in a rural or remote area, and do not actively engage in a hobby or other activity.

If you are feeling alone and stressed, don't be too hard on yourself – research has shown that many caregivers find it difficult to make new friends and maintain friendships. Many caregivers suffer from isolation, which can often lead to depression. But you don't have to suffer, there are actions you can take to feel more connected and reduce your stress:

- Ask for and try to get help
- Focus on the good and the difference you are making
- Set realistic goals
- Find and connect with caregiving resources in your community
- Set personal health goals
- Engage in self care
- Outdoor activities – go on a hike or just sit in a nice park for a while
- Physical activities – get active!
- Stay in touch with close friends and relatives
- Check out online support groups

Parents and caregivers of individuals with MWS have special needs, and clearly, more research is needed to better support them. If you have information or advice you would like to share with other caregivers of individuals with MWS, please contact us, we would love to hear from you!

MWS Matrix Portal

MOWAT-WILSON SYNDROME FOUNDATION Matrix

- The MWS Matrix Portal is up and running! This portal is available only for MWS families. The MWS Foundation in partnership with Across Healthcare have created a customized health records portal for MWS patients to help patients and caregivers easily capture, track, and share information related to medications, symptoms, activities, and electronic health records. The MWS Matrix Portal includes functions within the following categories:
- Medications: Here, you can create a log of all medications, dosages, and refill dates. Text and email alerts can be set up for medication administration and refill dates.
- Symptoms and Activities: Allows patients and caregivers to customize any symptoms or activities they want to track such as seizure activity, doctor appointments, medication changes, or sleep patterns. A graph is then created of multiple activities and symptoms to look for possible correlations.
- Journals: Allows for the creation of journals to easily keep a written narrative of important events, such as changes in eating, GI issues, physical therapy feedback, and doctor visits.
- Documents: Allows documents to be uploaded so they are easily accessible and in one place.
- Message Center: Allows for two-way communication with the MWS Foundation.
- Resource Center: Allows the MWS Foundation to make important documents and forms easily accessible. An example might be a document explaining MWS for a new health care provider, therapist, or teacher.

Have questions? Need help? Want to learn more? Keep an eye out for future newsletters with information about tutorials, webinars, and more!

Social Media

MWSF Facebook page:
https://www.facebook.com/mowatwilsonsyndromefoundation

MWSF LinkedIn Profile
https://www.linkedin.com/company/mowat-wilson-syndrome-foundation

MWSF Instagram Page
https://www.instagram.com/mowatwilson/

MWSF TikTok page
https://www.tiktok.com/@mwsfoundation?lang=en

Other MWS Community Facebook pages

Mowat-Wilson Syndrome Family
https://www.facebook.com/groups/56710348938

Life with Mowat-Wilson Syndrome
https://www.facebook.com/groups/9769306594

Mowat-Wilson Syndrome Community
https://www.facebook.com/groups/mowatwilsonsyndromecommunity

The mission of The Mowat-Wilson Syndrome Foundation is to enhance the lives of people affected by Mowat-Wilson Syndrome by providing family support, raising awareness, and supporting research and education.

Values

Compassion, Respect, Collaboration, Unity, Perseverance, Love, Dedication,
Honesty, & Integrity

Statement of Commitment to Diversity, Equity and Inclusion

The MWSF highly prioritizes fostering and maintaining an environment where diversity and inclusion are valued and achieved.

We strongly believe that everyone should be treated fairly. We greatly value all the diversity among MWS families and our staff, researchers, partners, vendors, volunteers, and other stakeholders. Through inclusion, we show our commitment to fairness and diversity. We greatly value the perspectives, abilities and identities of all our stakeholders. We highly respect and promote diversity in all its forms, including race, religion, color, ethnicity, national origin, socioeconomic status, ability, military service, age, faith, gender identity, sexual orientation, and personal appearance.

We are committed to being inclusive, respectful of each other, and honoring the right to be different. The experiences and perspectives of a diverse and equitable workforce and community increase the meaningfulness of our collaborations. We are committed to engaging in practices that support diversity, equity, and inclusion.

Members of the Mowat-Wilson Syndrome Foundation

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