MOWAT-WILSON SYNDROME FOUNDATION®

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.

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 www.mowat-wilson.org where you will find the latest information on MWS, past and upcoming MWS events, the MWS newsletter, and the MWS Patient Registry.

The MWS Patient Registry is used by doctors and researchers to learn more about MWS, which in turn helps us all learn more about this syndrome. Please see the Registry Information page included in this folder. We would like to encourage all families to go to the MWS Patient Registry and fill out all the surveys. You will be notified of the opportunity to fill out new surveys as they are developed.

We also 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, make a donation to the Foundation, join a Foundation committee, 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, specialists,and when you have emergency room or urgent care visits. Here are a few other suggestions that have been helpful to board members along the way.

Keep a list of all doctors, all medications, a history of seizures, and all diagnoses in the car for emergencies.

Keep a list at home of all doctors and therapists, their addresses and phone numbers, a list of people to call in an emergency, and the closest hospital with an emergency room.

Keep a portable notebook with all the pertinent information, as well as a seizure history, a medication history, and a history of all tests performed with results that you can carry with you if wanted or needed.

These are just 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 is made up of MWS family members who are truly invested in the mission of the Foundation and are all volunteers. Welcome and we hope to meet you at an event!

Sincerely,
The Mowat-Wilson Syndrome Foundation Board of Directors

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MWS PATIENT REGISTRY

Because we don't know what's possible

SHARE YOUR DATA

MOWAT-WILSON SYNDROME FOUNDATION®

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 Mowat-Wilson syndrome 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 Mowat-Wilson syndrome 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.

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)

Although many different medical issues have been associated with Mowat-Wilson syndrome, not every individual with this condition has all of these features.

Please review the need for specialists including genetics, neurology, cardiology, gastroenterology, nephrology as well as therapies including physical, occupational, and speech therapy.

MWS is caused by mutations in the ZEB2 gene. The ZEB2 gene provides instructions for making a protein that plays a critical role in the formation of many organs and tissues before birth. This protein is a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of particular genes. Researchers believe that the ZEB2 protein is involved in the development of tissues that give rise to the nervous system, digestive tract, facial features, heart, and other organs.

Mowat-Wilson syndrome 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 Mowat-Wilson syndrome.

Mowat-Wilson Syndrome Foundation Announces Award Grant for Establishment of Patient Registry

LAS VEGAS, NV - 10/07/2016 (PRESS RELEASE JET)

The Mowat-Wilson Syndrome Foundation is pleased to announce it was recently awarded a grant through Genetic Alliance, a leading nonprofit health advocacy organization, to establish a patient registry for individuals that have been diagnosed with Mowat-Wilson Syndrome (MWS). The grant will allow the Foundation to use Genetic Alliance's Platform for Engaging Everyone Responsibly (PEER) to build and house the registry.

The registry will facilitate the capture of key medical, genetic, treatment and demographic data for MWS patients. This in turn will allow expert clinical researchers to extract important information, study genotype correlations to clinical outcomes and it will aid physicians who care for individuals with MWS.

"As is true for many rare genetic conditions, our ability to collect clinically relevant information that can help physicians provide excellent patient care is limited because each provider has only a small number of patients with the condition," said Dr. Margaret Adam, Associate Professor of Pediatrics at the University of Washington Medical Center and member of the Mowat-Wilson Syndrome Foundation medical advisory board. "To further our knowledge of the breadth of MWS requires engagement of the involved community and a platform on which information can be securely and accurately entered and curated," she said.

An important feature of PEER is that it is highly customizable and parents or guardians of MWS patients can set their own sharing, privacy and data access preferences.

"The use of personalized sharing, privacy, and data access preferences is an important feature of PEER," said Sharon Terry, president and CEO of Genetic Alliance. "PEER enables cross-condition research where community is front and center: each group using the platform customizes their own portal to the registry, while connecting to other communities' portals on the system's backend. This empowers participants to contribute to research questions that matter to them, and simultaneously share their data more broadly – under data sharing and access permissions that they choose. We're thrilled to partner with MWSF in this vision, and look forward to seeing them launch!"

The Foundation hopes to have the registry up and running by October 15, 2016 and is aiming to have 100 participants engaged with the registry within the first year.

MWS is a recently discovered syndrome (defined in 1998) and much is still being learned about the physical, behavioral and developmental issues associated with this rare genetic disorder. Major signs include distinctive facial features, intellectual disability, delayed development, intestinal disorders, seizures, congenital heart disease, agenesis of the corpus callosum, male genital abnormalities (hypospadias), and major expressive language difficulty. Most Mowat-Wilson children are non-verbal and experience delayed development of motor skills such as sitting, standing, and walking. Despite their many difficulties, MWS children typically have friendly and happy personalities, though they will require intense medical attention and personal care throughout their entire lives.

To learn more about MWS visit www.mowat-wilson.org.

For more information on PEER visit: http://peerplatform.org.

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