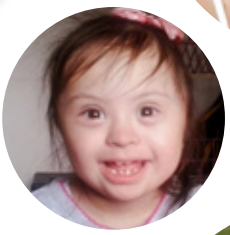
International Mosaic Down Syndrome Association

Uniting Unique People in a Unique World

www.IMDSA.org



# Information for Families & Professionals

# What is IMDSA?

The International Mosaic Down Syndrome Association is a volunteer-based, non-profit organization designed to support any family or individual whose life has been touched by mosaic Down syndrome (mDs) through their continuous pursuit of research opportunities and by increasing awareness in the medical, educational and public communities throughout the world.

IMDSA offers a variety of services and programs to meet the needs of all persons interested in mDs. Some of these services and programs include:

- **Family Connect Program** that allows members to connect with other families that have a loved one with mDs.
- **Self-Advocate Leadership Team (SALT)** that allows self-advocates aged 16 and over to connect and discuss topics with the guidance of a group leader. Meetings are conducted through video conference calls six times a year.
- **New Family Welcome Kit** provides information to help answer questions individuals and/or family members may have about mDs and how to deal with this new diagnosis.
- **IMDSA Database** that keeps track of vital information about our families and individuals with mDs. This information aids in research and allows the Family Connect program to be successful.
- **Research and Retreat Weekend Conference** that allows families and self-advocates to connect with each other and attend educational workshops to help them support and advocate for their loved one with mDs (or to self-advocate). This conference also allows professionals interested in mDs (or Down syndrome without mosaicism) to conduct research with attendees at the Conference.

## Our Mission

IMDSA is designed to support any family or individual whose life has been touched by mosaic Down syndrome by continuously pursuing research opportunities and increasing awareness in the medical, educational, and public communities throughout the world.

# What is Mosaic Down syndrome?

Medically, this condition is known as mosaicism for trisomy 21. Throughout this document it will be referred to as mosaic Down syndrome or mDs. An individual with mDs has two or more types of cells; usually a percentage of cells with an extra copy of chromosome 21, as well as a percentage of cells that have the typical number (2 copies) of chromosomes 21 (unaffected). This condition occurs as a result of a chromosome sorting problem(s) that arise prior to and/or after conception and is no fault of the parents (they did not do anything to cause the chromosome sorting problem).

According to research, 2–4% of people in the Down syndrome community have mDs. However, this number may be an underestimate since it does not fully account for individuals who never receive a diagnosis or those who are initially diagnosed as having non-mosaic Down syndrome, with that diagnosis being refined to mDs following further studies.

- Average age of diagnosis is 1–4 years old
- Not restricted to any race, culture, or religion
- Physical characteristics show a wide range of variation and may be mild
- IQ levels are generally 10–30 points higher than those with non-mosaic Down syndrome
- Slightly delayed motor skills can be helped with therapy
- May require some degree of special education
- The Down Syndrome Medical Guidelines should be followed to ensure continued health (see tables in this brochure)
- The learning potential will vary between individuals, but some adults go to college, marry, and have children
- The risk for a person with mDs to have a child with Down syndrome will vary, depending on the percentage of reproductive cells that have an extra chromosome 21
- Research shows that siblings of those with Down syndrome/mDs are well adjusted and often pursue careers in health care, law, or education
- Translocation mosaicism can also occur. About 2–4% of all people with non-mosaic Down syndrome have a trisomic imbalance for chromosome 21 due to a translocation. Mosaicism for Down syndrome due to a translocation, or other structural chromosomal finding, is less commonly seen than mDs due to trisomic cells with three structurally typical chromosomes 21.

# Diagnosis: After Birth

To diagnose mDs, a genetics laboratory evaluates the individual's chromosomal make-up using either a conventional chromosome test (ideally, at least 30 cells should be counted), a fluorescence in situ hybridization test (many cytogenetic labs will score at least 100 cells), or a microarray test (typically, individual cells are not evaluated with this method, but it is sensitive for detecting mosaicism present in approximately 5%–10% or more of cells).

It is important to recognize that the proportion of trisomic cells can vary among tissue types. For example, there may be differences in the percentage of trisomic cells in the blood as compared to the skin, heart, or other organs. For this reason, one may wish to evaluate more than one type of tissue for diagnostic testing.

## Understanding Test Results/Karyotypes to Parents

Test results can be divided into sections. Our goal is to help you understand these results.

**1) Specimen type:** Some reports will indicate the type of specimen used for the test [e.g., blood cells or other tissues (such as skin cells)]. Because these cell types have different embryonic origins, they could have different percentages of cells with an extra chromosome 21. For example, a child with mDs may have an extra chromosome 21 in 60% of their skin cells but only 30% of their blood cells. Research studies are being done to determine what these different percentages mean.

**2) Metaphase Cells:** Metaphase occurs when cells are dividing. Since the DNA condenses at this stage, it is possible to count the number of chromosomes present.

**Metaphases Counted:** This value indicates how many cells were evaluated to determine chromosome number. This assessment does not include a band-by-band detailed analysis.

**Metaphases Analyzed:** This value indicates how many cells were evaluated in a band-by-band manner to look for small amounts of extra or missing genetic information.

**Metaphases karyotyped:** This metric indicates how many cells had pictures

(karyograms) prepared. A karyogram is a term that refers to an ordered arrangement of the chromosomes.

**3) Cultures:** The number of cultures shows that cells were grown in more than one culture vessel to ensure that the findings seen are representative of those present in the individual (and not artifact from the lab growth process). Typically, at least two independent culture vessels are used. Results from two or more cultures are more reliable than results from one.

**4) Banding techniques and resolution:** This information describes the methods that were used for the test.

**5) General testing interpretation information:** All of the organs (blood, heart, liver, pancreas, etc.) in our body are made up of cells. Each cell contains the genetic information (instruction manual) that our body needs to grow and develop. The genetic information is packaged into chromosomes. There are 23 pairs of chromosomes, and each pair contains a different set of genetic instructions. When most of us are conceived we get one copy of each chromosome from our mom in the egg and one copy of each chromosome from our dad in the sperm. Since each parent contributes 23 chromosomes, we wind up with a total of 46 chromosomes.

The first 22 pairs of chromosomes are the same in males and females. The sex chromosomes will differ between males and females. Most females have two X chromosomes and most males have one X and one Y chromosome.

Cytogeneticists use a shorthand method for describing chromosome results. The shorthand form for a typical female is: 46,XX. The number 46 indicates that there were 46 chromosomes present, and the XX indicates there were two X chromosomes observed. For most males, this shorthand form would be: 46,XY.

If your child has been diagnosed with Down syndrome, the karyotype could have an extra chromosome 21. If the child were a girl, it would be written as: 47,XX,+21 [indicating that this child has a total of 47 chromosomes, including two X chromosomes (which is the biological complement associated with female traits) and one extra copy of a chromosome 21].

Mosaicism is indicated by showing a karyotype result for the different types of cells that are present in the child's body. For people having mDs, most frequently one cell type has a typical chromosomal make-up and the other cell type has an extra chromosome 21. For example, your child may have received results that look something like this: 47,XX,+21/46,XX. In this case, the two different types of cells present in the individual are separated by a slash ( / ).

If this were a boy the results would look like: 47,XY,+21/46,XY

In some cases, the actual number of cells having each type of chromosomal make-up will be specified in the karyotype. For example: 47,XX,+21[15]/46,XX[6]. This means that 15 cells had an extra chromosome 21 and 6 cells had a typical chromosomal make-up.

## What does a mosaic karyotype mean for my child's development?

There is a wide range of outcomes for people with mDs. Unfortunately, we don't currently have a way to accurately predict what the outcome will be for each infant with mDs; just like we cannot predict the outcome of a baby who has a typical chromosomal make-up. The need for more information is why IMDSA strongly supports research so that we can better interpret test results and develop optimal health care and educational management approaches for people with mDs.

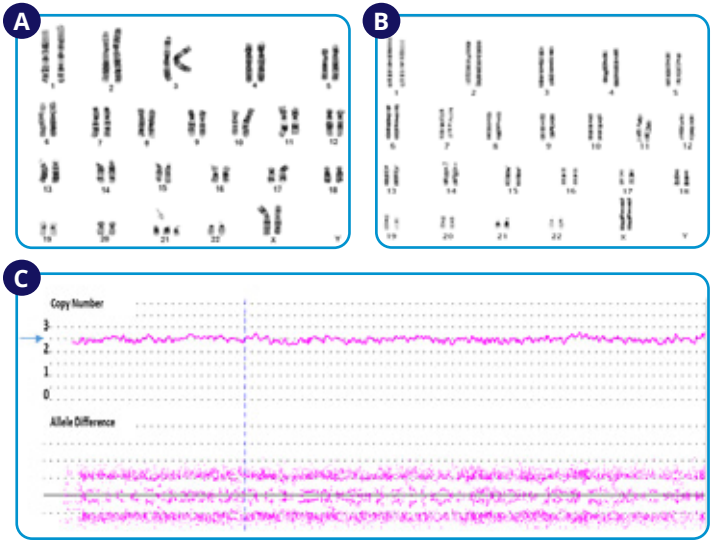
**6) Recurrence risk:** Many parents would like to know what the chances are of having another child with mDs. Some result summaries might indicate that the couple has an "increased risk." This statement is based on studies of children with non-mosaic Down syndrome. There are no large studies evaluating specifically the recurrence risk of mDs. However, the studies on non-mosaic Down syndrome indicate that the chance of having a second child with Down syndrome is about 1%. This is greater than the population risk of about 1 in 800.

**7) Referral for genetic counseling:** Many reports will recommend that the patient receive genetic counseling or an additional genetic evaluation. The genetics team can help to coordinate care for individuals with genetic conditions. The team will tell you what is known about the causes of mDs and explain what you can expect based on current research. They will also make sure that you have all of the referrals you need to ensure your child is getting the right medical care and early intervention services. Genetic counselors can be a valuable resource to provide information about support groups and answer questions you might have about mDs.

IMDSA strongly supports research so that we can better interpret test results and develop optimal health care and educational management approaches for people with mDs.

# Diagnosis: Before Birth
## (Prenatal)

Some women/couples elect to have information about the chromosomal make-up of an embryo (up to the 8th week of gestation) or fetus (8th week of gestation to term) while they are pregnant. This information can be used to enable them to make decisions about the management of the pregnancy or decisions about continuing the pregnancy. One approach for determining if a pregnancy involves a trisomic conceptus is a screening assay called Non-Invasive Prenatal Testing (abbreviated as NIPT). This screening test (which might also be called a cell-free DNA screening test) is performed by taking a blood specimen from a pregnant woman (at least 10 weeks gestation). The result of this screening test identifies pregnancies that are at an increased risk for having a child with Down syndrome (or other chromosomal findings). However, since it is not a diagnostic test, it is recommended that a woman who receives a positive NIPT screen result consider a follow-up prenatal diagnostic test to determine if the fetus does indeed have trisomy 21 (or mosaicism for trisomy 21 [or another chromosomal finding]) to aid them in making decisions regarding the pregnancy. Two types of diagnostic tests are available: (1) chorionic villus sampling (abbreviated as CVS), which is performed during the first trimester (typically 11 to 14 weeks gestation); or (2) amniocentesis, which is performed during the second trimester (typically 15 to 20 weeks gestation). It is recommended that a woman/couple considering prenatal screening/testing receive genetic counseling to aid them in understanding the results of the tests. Also, it is important that a woman/couple who has received a result that is positive for Down syndrome or mDs gain accurate information about these conditions. Genetic counselors can provide up-to-date information. At some centers, they can also connect the woman/couple with parents of children who have Down syndrome or mDs to provide opportunities for them to ask questions about life experiences related to these conditions and enable her/them to make this highly personal decision based on accurate information.




*Most people with mosaic Down syndrome have 2 types of cells: **(A)** those with trisomy for chromosome21 (arrow); and **(B)** those with typical chromosome number (two for each chromosome). Another test that can be used to identify mosiacism is called a microarray test **(C)**. This patient has a copy number that falls between 2 and 3 (in this case 2.5) indicating that this individual has mosiacism. The Allele difference pattern also shows a pattern consistent with mosiacism for trisomy 21.*

# Developmental Milestones

As the parent of a baby or toddler with mDs, you may be concerned about their developmental growth. It is important to remember that your baby or toddler will reach these milestones, but it may take a bit longer than the timelines noted for children with typical chromosomal make-ups. Sometimes your baby or toddler may need a little extra help to develop the skills they need to reach these steps in development.

To help your baby or toddler, a physical, occupational, and/or speech therapist may be needed. You can contact your physician or government offices to find out about the services available in your area. With you and your therapist's help, your child will reach these important milestones.

In the following table, we provide a guide of average timelines for milestone attainment that has been observed in children with mDs. However, not all babies are exactly alike! Each one will reach his or her milestones in his or her own time. Your child may reach milestones sooner or later than the range noted in the table. It is provided to help you know if your baby may need a little extra help in certain areas of development. It is important to remember that even when it doesn't look like your baby is progressing, they are always working on acquiring new skills; some are just more visible than others.

One of the most common concerns among parents of children with mDs is speech delays. Although many children take longer to acquire communication skills, the majority of individuals with mDs develop speech patterns that are readily understood.

## Average Milestone Attainment Times Reported by Parents for Their Children Having mDs

| | |
|---|---|
| **Roll Over** | **4 – 5 months** |
| **Sit Without Support** | **8 – 9 months** |
| **Crawl** | **11 months** |
| **Drink From Cup Unassisted** | **17 months** |
| **Walk Unassisted** | **20 – 21 months** |
| **Walk Up Stairs** | **24 – 25 months** |

# Healthcare Guidelines

## Birth – Five Years Old

Children with mDs have the same healthcare needs as any child. However, because individuals with mDs have a portion of their cells with an extra chromosome 21, they can develop many of the health problems associated with "full" (non-mosaic) trisomy 21. Therefore, it is important that they get the same tests and evaluations that are recommended for individuals with Down syndrome.

During your child's routine evaluations, the doctor will do all the standard exams that they do for people of his/her age, but they may also complete some extra testing. Since individuals with mDs are sometimes smaller in size, your doctor may compare your child's growth with the growth chart for children having Down syndrome. Additional tests for children from birth to five years of age may include an echocardiogram to assess potential heart abnormalities, a thyroid screening, and a neck X-ray. Please refer to the recommended Health Care Guidelines for children with Down syndrome to help you know when your child may need these exams (also see the pediatric health care guideline table in this brochure).

Although there are many health problems associated with mDs, this does not mean that your child will have all of these conditions. It simply means that since they are at risk, annual check-ups are necessary to ensure your child stays healthy.

Recently, health care guidelines have also been developed for adults who have Down syndrome. Akin to the scenario for children, adults with mDs are unlikely to have all of the conditions that have been associated with Down syndrome, but it is important that they follow these guidelines to help keep them healthy or identify potential health issues as soon as possible (see table summary of adult health guidelines in this brochure).

# Social and Emotional Health

Children and adults with mDs have the same social and emotional needs as people without mDs or Down syndrome. They experience happiness, joy, sadness, and fear. People with Down syndrome tend to highly value social interaction, and this is true for people with mDs as well. Like all of us, they need social connection, acceptance, inclusion, and self-determination to do well and feel well.

Some research suggests that people with mDs and/or Down syndrome may be more prone to symptoms of anxiety and depression. This is especially true for people who have experienced social difficulties, such as feeling left-out, teased, or bullied. Supporting healthy social skills and healthy social relationships is an important part of supporting emotional health. People with mDs and Down syndrome also tend to use "self-talk." Self-talk is something we all do, and for most people self-talk is an internal dialogue. Realistic and positive self-talk generally helps us, whereas negative or unrealistic self-talk can lead to anxiety and depression. Many people with mDs and Down syndrome say their self-talk out loud. We can help to support their mental health by encouraging coping self-talk (e.g., "I can't do my usual routine, but I can handle it.").

## Here are some other strategies for promoting healthy social and emotional development:

**Childhood.** Use positive behavior supports to develop consistent routines and rules. Help your child develop an emotion vocabulary by labeling their emotions, labeling the emotions of others, reading books about emotions, and playing emotion games (e.g., emotions charades). These skills are important for understanding and communicating emotional needs in an appropriate way. Have frequent supervised playdates with other children to build social skills. Children with developmental delays need lots of opportunities to practice. Strive to have playdates with children with typical development and children with similar special needs.

**Adolescence.** Social relationships become very important during adolescence. The results of research suggests that children need at least one close friend. Participation in sports or other social activities can help give adolescents an opportunity to find and develop meaningful friendships and develop special talents and skills. Continue to talk openly about emotions and coping strategies. Your adolescent will likely have

questions about dating and sexuality. Encourage discussions about healthy romantic relationships, healthy touch, and healthy boundaries.

**Adulthood.** For adults, feeling like one is able to be self-determined and make a contribution to one's community are important for well-being. Some adults with mDs may need help identifying potential career opportunities. They may also need encouragement to be as independent as possible, whether that means living independently, living independently with support, or living at home. They may also benefit from continued support to develop meaningful social and romantic relationships.

**Seeking Mental Health Care.** If you have questions or concerns about your child's emotional or social health, talk with your child's primary care provider and/or a licensed mental health care provider. It is important to note that some medical problems that are not uncommon in people with mDs can cause emotional symptoms, such as a thyroid imbalance or sleep apnea, so these should be checked. If you or your child's doctor believe that he/she may benefit from counseling, look for therapists with expertise in cognitive behavioral therapy (CBT). CBT has been shown to be effective for a wide range of emotional, behavioral, and social problems. It has also been shown to be effective with people with developmental delays or mild to moderate intellectual disabilities.

Feeling like one is able to be self-determined and make a contribution to one's community are important for well-being.

*Family Connect Day Outing, Research and Retreat Weekend. Cincinnati, OH, 2018*

# Chart

## Adapted Health Supervision Guidelines for Children with Down syndrome (mosaic or non-mosaic).*

| Health Review Activity/Concern |
|---|
| Parent-to-parent contact; support groups; books and pamphlets |
| Physical exam for trisomy 21 or mDs |
| Chromosome analysis to confirm diagnosis |
| Genetic counseling to discuss recurrence risk |
| Echocardiagram |
| Newborn hearing screen and follow-up |
| Radiographic swallowing assessment if marked low muscle tone, feeding issues |
| Eye exam for cataracts |
| Exam for assessment for duodenal or anorectal atresia |
| Discuss dental issues (delays in eruption; fewer teeth) |
| If constipation; evaluate diet, muscle tone, hypothyroidism, GI malformation, Hirschsprung |
| Hemoglobin, CRP and ferritin or CHr tests |
| Hemoglobin |
| TSH (may be part of newborn screen) |
| Discuss cervical spine positioning, especially for anesthesia, surgical, or radiologic procedures |
| Review signs and symptoms of myopathy |
| Instruct to contact physician for change in gait, arm or hand uses, bowel or bladder functions, neck pain, head tilt, or new-onset weakness |
| Review risks for contact sports, trampoline |
| Audiology evaluation |
| If normal hearing known; behavioral audiogram |
| Assess for obstructive sleep apnea |
| Sleep study |
| Eye exam: Asses for cataracts; strabismus; nystagmus |
| Follow-up eye exams with ophthalmologist experienced with Down syndrome |
| Check for symptoms of celiac disease; if present, follow-up as indicated |
| Early intervention, physical, occupational, and speech therapy |
| Discuss self-help skill, learning behaviors, wandering off, social progress |
| Changes related to puberty, gynecologic care in pubescent females |
| Discuss sexual development/behaviors, contraception, recurrence risk for offspring |
| If congenital heart disease, monitor for symptoms of congestive heart failure |

**Adapted from: Bull MJ and the Committee on Genetics (2011). Clinical Report – Health Supervision for Children with Down Syndrome. Pediatrics 128 (2): 393-406 (Note: these guidelines were re-affirmed in 2014). Consult your child's physician for the full guideline list, which can be accessed at: (https://pediatrics-aappublications-org.proxy.library.vcu.edu/lens/pediatrics/128/2/393#).*

Complete only if not done previously

Do once at this age

HM = Health maintenance

| Birth – 1 mo | 1mo – 1 yr | 1 – 5 yr | 5 – 13 yr | 13 – 21 yr |
|---|---|---|---|---|
| X | X | X | | |
| X | | | | |
| X | X | X | | |
| | X | X | | |
| X | | | | |
| X | | | | |
| X | X | | | |
| X | X | | | |
| X | | | | |
| | X | | | |
| Any visit | | | | |
| | | Annually | | |
| | | | | Annually |
| X | 6 & 12 mo | Annually | | |
| All health maintenance (HM) visits | | | | |
| All HM visits | | | | |
| | | Biennially (every other year) | | |
| | | All HM visits | | |
| | 6 mo | | | |
| | | Annually | | |
| | X | All HM visits | | |
| | | By age 4 yrs | | |
| | X | | | |
| | | Annually | Every 2 yrs | Every 3 yrs |
| | | All HM visits | | |
| | | All HM visits | | |
| | | | All HM visits | |
| | | | | HM visit |
| | | | | HM visit |
| | All Visits | | | |

Complete only if not done previously
HM = Health maintenance

Do once at this age

# Chart

## Adapted Health Supervision Guidelines for Adults with Down syndrome (mosaic & non-mosaic).*

| Health Activity/Concern |
| --- |
| Follow general population adult health guidelines. |
| |
| Discuss behavioral, functional, adaptive, and psychosocial factors |
| Symptoms of a mental health disorder are present |
| |
| Discuss behavioral patterns related to dementia |
| Symptoms of loss of function and/or memory loss suggestive of possible dementia |
| |
| Discuss risks for diabetes |
| Adults who are overweight |
| |
| If no history of atherosclerotic cardiovascular disease |
| Adults with a history of congenital heart disease |
| |
| Discuss healthy diet, regular exercise, and calorie management |
| |
| Discuss risks for atlantoaxial instability |
| |
| Discuss risks for osteoporosis |
| Symptoms of fragility fracture |
| |
| Discuss risk for hypothyroidism |
| |
| Discuss risk for celiac disease |

**Adapted from: Tsou, et al (2020). Medical Care of Adults with Down Syndrome: A Clinical Guideline JAMA 324(15): 1543 – 1556 and Global Down Syndrome Foundation (https://www.globaldownsyndrome.org/wp-content/uploads/2020/10/Global-Down-Syndrome-Foundation-Medical-Care-Guidelines-for-Adults-with-Down-Syndrome-Checklist-v.1-10-20-2020.pdf.) Consult your (self-advocates) or your adult child's (parents) physician for the full guideline list.*

| | 21 – 29 yr | 30 – 39 yr | 40 – 49 yr | 50 – 59 yr | 60+ yr |
|---|---|---|---|---|---|
| | **All health maintenance visits (In addition, note the following areas of health focus).** | | | | |
| **Behavior** | | | | | |
| | **All Health Maintenance visits** | | | | |
| | When symptoms/concerns for a mental health disorder arise, evaluate for medical conditions and schedule appointment with clinician familiar with Down syndrome (medical professional should use the DSM-5 or DM-ID-2 guidelines) | | | | |
| **Dementia** | | | | | |
| | | | **Interview primary caregiver(s) to assess changes from baseline function annually beginning at age 40** | | |
| | Caution is indicated when diagnosing Alzheimer's Type Dementia in people less than age 40 (include assessment of other health conditions) | | Decline in the six domains per the National Task Group-Early Detection Screen for Dementia should be used to identify early-stage, age-related Alzheimer's-type dementia and/or a potentially reversible condition | | |
| **Type 2 Diabetes** | | | | | |
| | | If asymptomatic, screen every 3 years | | | |
| | Screen every 2 to 3 years (screen using HbA1c or fasting plasma glucose) | | | | |
| **Cardiac** | | | | | |
| | | | **The use of statin therapy should be assessed every 5 years (starting at age 40) & using a 10-year risk calculator like general population** | | |
| | Given the increased risk for having a stroke, a periodic cardiac evaluation and monitoring plan should be reviewed by a cardiologist | | | | |
| **Obsesity** | | | | | |
| | **All health maintenance visits** | | | | |
| **Atlantoaxial Instability** | | | | | |
| | **All Health Maintenance visits; Annual screening should be completed in adults based on signs/ symptoms of cervical myelopathy using targeted history and physical exam (routine cervical spine x-rays should not be used to screen asymptomatic individuals)** | | | | |
| **Osteoporosis** | | | | | |
| | There is insufficient evidence to recommend for or against applying general population screening guidelines. At this time, good clinical practice supports a shared decision-making approach | | | | |
| | Should be evaluated for secondary causes of osteoporosis, including screening for hyperthyroidism, celiac disease, vitamin D deficiency, hyperparathyroidism, and medications associated with adverse effect on bone health. | | | | |
| **Thyroid** | | | | | |
| | **Annual health maintenance visits; Screening should be performed every 1 to 2 years using a serum thyroid-stimulating hormone (TSH) test** | | | | |
| **Celiac Disease** | | | | | |
| | **Annual health maintenance visits (Assessments include targeted history, physical exam, and clinical judgement)** | | | | |

# Speech and Feeding

- People with mDs show great variability in their speech intelligibility
- One of the most common speech problems, called apraxia, occurs when an individual has difficulty planning and producing the mouth movements necessary for speech or articulation
- Other areas of difficulty that may require treatment from a speech and language pathologist include:
    - Feeding/swallowing (Dysphagia)
    - Understanding spoken language and following directions (Receptive Language)
    - Expressing wants, needs, and ideas using spoken language (Expressive Language)
    - Producing fluent speech (Stuttering)
    - Using and understanding elements of syntax or grammar
    - Using speech for pragmatic or social functions, such as retelling stories, asking for help, or engaging in conversations with peers
- Studies are currently underway to better understand the causes of speech variations and to identify correlations with the proportion of trisomic cells

Apraxia occurs when an individual has difficulty planning and producing the mouth movements necessary for speech or articulation.

# Motor Skills

Development in children with mDs is varied, but certain challenges have been consistently identified.

## Gross Motor Skills

- Low muscle tone at birth may make it difficult for the child to lift his/her head
- Children are often late to sit up, roll over or walk
- Sometimes individuals have balance and coordination difficulties
- Joints may be less stable, which may cause differences in their gait
- Low muscle tone and lack of strength may result in the person having a rounded sitting posture

## Fine Motor Skills

- Children with mDs often have short fingers and joint instability
- Children with mDs may have difficulty with bilateral coordination, in-hand manipulation skills, and hand-eye coordination
- Children with mDs may have difficulty learning to write legibly, managing clothing fasteners, and coordinating visual motor skills
- Some children with mDs may also have difficulty coloring, cutting, and manipulating materials

# Learning

Early intervention is often crucial to help a child with mDs/Ds meet their developmental milestones, though not every child will need special services or academic assistance.

- In the United States, mDs/Ds is *not* one of the 13 disability categories outlined in the Individuals with Disabilities in Education Act (IDEA). However, a student may qualify for an Individualized Education Plan (IEP) under another category (for example, specific learning disability or speech impairment).
- Open communication about the IEP will help achieve success in the classroom. Supportive communication exchanges between parents and educators will help to create a "team" approach wherein parents and educators feel like they are on the same side in trying to help the student with mDs/Ds.
- Individuals with mDs often struggle with math and science. Cue cards can be useful for difficult concepts, such as math formulas or technical vocabulary.
- Students with mDs may have some difficulty understanding instructions—especially those involving multiple steps. It is important to be in tune with the student's understanding of instructions. Paraphrasing, prompts, and cues may be necessary. Also, several students with mDs tend to be visual learners, so providing pictorial directions (rather than verbal or written directions) can help enhance their understanding of a task (or multiple steps in a complex task). Also, since some people with mDs may have poor vision, it may be helpful to provide them with bold, enlarged images or materials.
- Other helpful classroom accommodations include extended time, modified assignments/tests, and the use of technology (calculator for math, word processor with spelling and grammar check for written assignments).
- Self-esteem is very important for the individual with mDs. Ensure that a zero-tolerance to bullying is in place throughout the school day.

During high school, the student may receive transitional services that include a paid or unpaid job.

# Delivering the News About the Diagnosis

Family members receiving a diagnosis of mDs for a child are likely to experience many different thoughts and emotions. This can be a stressful, overwhelming time. Some parents of children with mDs report feeling supported when their health care providers (and/or friends/extended family members) do the following:

- Congratulate the family on the birth of their child with mDs.
- Find something positive to say about the individual who has been diagnosed with mDs.
- Interact with the child so the families know you are comfortable with them.
- Provide hope; times are changing and people born today with mDs/Ds have many opportunities for a rewarding life.
- Remove parental liability. Explain to the parents that nothing they did caused their child to have mDs.
- Use "people first language."
- Some families feel they are unable to deal with a child who has special needs. If the family does not seem to be able to cope with the diagnosis, be prepared to offer them options for adoption.
- Provide accurate information about mDs.
- Provide information about IMDSA so families know support is available. Some families may also benefit from local support groups for non-mosaic Down syndrome.
- Many parents would like to know what the chances are of having another child with mDs. While there are no large studies that specifically report the risk for recurrence in mDs, the recurrence risk associated with Down syndrome indicates that the chance of having a second child with this condition is about 1% for families having a child with an entire extra chromosome. This is greater than the population risk of about 1 in 800. Families having a child with mDs (or non-mosaic Down syndrome) due to a structural chromosome change (such as a translocation) will have a different risk that will be explained by their genetic counselor. Maternal age should also be a consideration when counseling for chromosomal conditions.
- Many families benefit from a referral to a genetic counselor.

# Clinical Features

- The clinical findings of mDs are similar to those seen in individuals with Down syndrome, although they may be present in a milder form.
- There is a great deal of variability in the clinical findings of individuals having Down syndrome, but this range is even broader in people having mDs.
- Characteristic facial traits include upward slanting eyes with epicanthal folds (fold of skin in the inner corner of the eye), a flat nasal bridge, tongue thrusting, and small ears.
- Common medical problems include cardiac defects, hearing and vision problems, and respiratory and/or thyroid conditions.
- Individuals with mDs/Down syndrome have an increased risk for developing leukemia.
- Some people with mDs will have cognitive impairment in the mild to moderate range and benefit from early intervention services soon after birth to address developmental and speech delays.
- While there is a correlation between the percentage of cells with trisomy 21 in the peripheral blood or buccal mucosa (cheek) cells and the number of clinical findings observed in the child/individual, this correlation is not perfect or predictive.
- Children with mDs need immunizations and well-child care similar to the strategies followed for children without trisomy 21.

**"Health Guidelines for Individuals with Down Syndrome," published by the American Academy of Pediatrics committee on genetics is a useful resource for pediatricians.**

**Find these guidelines at http://imdsa.org under Information > Information Packets > Down syndrome Medical Checklist (an abbreviated version is presented in the table in this booklet)**

*https://pediatrics-aappublications-org.proxy.library.vcu.edu/lens/pediatrics/128/2/393#*

# Top Questions Asked by Families

Families throughout the world come to IMDSA daily to ask questions concerning their child's diagnosis. We hope that these FAQs help you better understand your child or clients/patients/students, depending on your role in the life of a person with mDs.

### Q) How is mDs going to affect my child's development?

**A)** Every child is different. We cannot predict the outcome for an infant with mDs, but it is important to remember that we also cannot predict the outcome for a child who has a typical chromosome make-up. A diagnosis of mDs provides a list of health and development concerns that could arise, but most people with mDs (or non-mosaic Down syndrome) will not have all (or even most) of these health or learning concerns. It is important to identify areas of interest and strength/weaknesses for your child and provide them with opportunities to improve and succeed.

### Q) Will my child have more medical problems?

**A)** Your child is at a higher risk for the medical conditions associated with Down syndrome. However, this does not mean that your child will develop all of these conditions. With proper medical intervention outlined in the Health Guidelines for Down syndrome your child can live a healthier life.

*Self-advocates with mDs who attended an IMDSA Research and Retreat Weekend. Colorado Springs, CO, 2017*

**Q) Do people with mosaic Down syndrome have fewer learning/developmental problems than people with non-mosaic Down syndrome? If my child was diagnosed with non-mosaic Down syndrome, but demonstrates strong learning skills does that mean he or she might have mosaicism?**

**A)** People with mosaic Down syndrome, and non-mosaic Down syndrome, show a wide range of abilities, just like people with typical chromosomal make-ups show a wide range of abilities. Overall, standardized testing results (e.g. IQ testing) for people with mosaic Down syndrome tend to fall on a continuum in which their scores are lower than those of their siblings, but higher than people with non-mosaic Down syndrome. However, learning skills and other talents are influenced by many factors (genes other than those located on chromosome 21, environmental influences, etc.). On a case-by-case basis, some individuals with mosaicism may have more learning problems than individuals with non-mosaic Down syndrome. Also, many people with non-mosaic Down syndrome have very strong skills; far beyond those sometimes described in older literature. Working with educators, occupational therapist, and other learning and care providers can help to identify opportunities for growth, as well as areas of strength/talent, for each child or adult. This knowledge can be used to optimize performance and success in school, the workplace, and social interactions.

**Q) What is the life expectancy for a person with mosaic Down syndrome?**

**A)** Currently we need further research to know the answer to this question. However, the oldest living person with mosaic Down syndrome was noted to be 83 yrs old. (Brian Chicoine, MD & Dennis McGuire, PhD. 1997.) With current medical treatments, most individuals should live long fulfilling lives.

**Q) Will my child have children when he/she grows up?**

**A)** Females with mDs usually are fertile. Also, while males with non-mosaic Down syndrome tend to be infertile, males with mDs have fathered children (so they can be fertile). Note that a person who has mDs does have an increased risk for having a child with non-mosaic Down syndrome, but they could also have a child with typical chromosomes. When they reach reproductive age, people with mDs (and/or their family members) may benefit from genetic counseling to better understand their risk for having a child with Down syndrome (which will vary from person to person and depends on the percentage of trisomic cells that are present in their reproductive cells).

**Q) What can I do to help my child succeed?**

**A)** Early intervention and medical check-ups are important. However, the best way for you to help your child succeed is to treat him/her just like you would any other child. Expect the very best from your child and he/she will give you his/her very best!

# People First Language

In today's society there is a politically correct term for every facet of life. The same goes when discussing a person having mDs. Some people are confused about the pronunciation of this chromosomal condition. Several years ago the condition was pronounced "Down's" syndrome (named after Dr. John Langdon Down who first described this condition). However, since Dr. Down did not have trisomy 21, the apostrophe is not accurate. As a result, this condition is currently described simply as "Down" syndrome.

**When speaking about a person with mDs . . .**

- Use people first language
- Always put the person first in word and thought
- Be accurate and non-judgmental
- Emphasize abilities

Do not say the person "is Mosaic" or "is Down syndrome." Think of a person who has high blood pressure—you would not say the person "is high blood pressure"!

In the same respect, you would not say "the mosaic Down syndrome person." The recommended way to discuss a person with this condition is "The person with mosaic Down syndrome" or the person having mosaic Down syndrome (or Down syndrome). But, please keep in mind, that this is only a medical diagnosis for that person, not who the person is. This applies to anyone with a genetic condition or ANY health problems. People-first language will help the individual feel much more comfortable around you, and it is quite simple to use!

People-first language will help the individual feel much more comfortable around you, and it is quite simple to use!

# Additional Resources

The following websites provide additional information about Down syndrome and may be helpful:

**The DS-Connect.nih.govTM registry** is an especially helpful site. Registering at this site is similar to providing information for the census. It helps to identify how many people have mDs or non-mosaic Down syndrome. In turn, this information is useful to researchers designing studies for treatments related to Down syndrome. It is also helpful to researchers who are trying to explain the importance of gaining insight about Down syndrome or mDs. This site also provides useful information about health care updates and clinical trial opportunities.
**Website: dsconnect.nih.gov**

**DSC2U** is a website that was created by Dr. Brian Skotko, MD, MPP, and the clinical and research teams from the Down Syndrome Program at Massachusetts General Hospital, a not-for-profit academic hospital affiliated with Harvard Medical School. This site was created to "help families get up-to date, personalized health and wellness information based on national guidelines and expert consensus, for a person with Down syndrome" (Chung, et al., (2021) Genetics in Medicine 23:163-173).
**Website: www.dsc2u.org**

**Global Down Syndrome Foundation** is a public non-profit 501(c)(3) organization. Their goal is to significantly improve the lives of people with Down syndrome through Research, Medical Care, Education, and Advocacy. They work to educate governments, educational organizations, and society in order to affect legislative and social changes so that every person with Down syndrome has an equitable chance at a satisfying life. The IMDSA has partnered with Global Down Syndrome to expand the success and outreach of both organizations.
**Website: www.globaldownsyndrome.org**

**The National Down Syndrome Congress** is dedicated to an improved world for individuals with Down syndrome. They are the leading national resource of support and information for anyone touched by or seeking to learn about Down syndrome. They strive to educate, advocate, empower, and inspire. This group was especially helpful for connecting people with mDs during the early years of the IMDSA development.
**Website: www.ndsccenter.org**

**The Down Syndrome Diagnosis Network** is the largest national organization supporting new and expectant parents with a Down syndrome diagnosis. The mission of the Down Syndrome Diagnosis Network is to connect, support, and provide accurate information to parents—and the medical professionals who serve them—from the time of diagnosis through age three, while fostering the opportunity for life long connections.
**Website: www.dsdiagnosisnetwork.org/**

**The Down Syndrome Adoption Network** has a mission to ensure that every child born with Down syndrome has the opportunity to grow up in a loving family.
**Website: www.ndsan.org**

# Advice from Adults with mDs

## Meet Rob

I graduated with a diploma from an Expeditionary Learning High School in 2009. Then I went on to graduate from a Community college in 2013 with six welding certificates. For two years during that time, I lived away from home in student housing in a pod of four separate rooms with eight students total. I started working at Distinctive Welding as a shop assistant/welder in August 2013 and have worked full time since then. During 2020/21, I was able to do more welding and installations. Do what makes you happy!

## Meet Felicia

A diagnosis is not something negative, despite what we have been expected to believe. Expect your child to do their best and teach them to advocate for themselves. Though others may think more or less of a diagnosis, to us, this is our normal; we do not need 'special treatment.' We need adjustments to be made to accommodate our learning differences, and to be treated like everyone else. We are all trying to live our best lives.

I am Felicia and I have mosaic Down Syndrome. I have been married for thirteen years and have two beautiful boys, who did not inherit my funky chromosomes! They are both full of energy and keep me busy! Presently, I am a stay-at-home mom. Last year, I completed Certificate IV in Education Support and I am in the process of looking for part time work as a Teacher's Aide, a job that involves working with children with disabilities in a school or pre-school setting to help them to achieve all that they can be.

# Anxiety, Depression, and Peer Relationships

## in People with Mosaic Down Syndrome

This study was conducted by Dr. Ruth Brown and Dr. Colleen Jackson-Cook of Virginia Commonwealth University. 25 people with mosaic Down syndrome (mDs) and 22 parents answered questionnaires at the IMDSA Research Retreat Weekends from 2013 to 2017 about symptoms of anxiety, depression, and peer relationship problems.

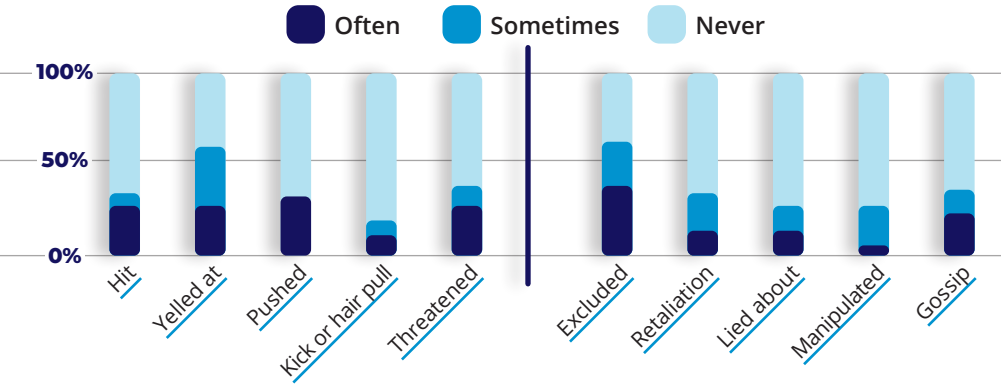
### **54%** Report Elevated Depression

*Using a self-report depression screen, 56% reported symptoms of depression above the recommended cut-off. This does not necessarily mean that 56% have a depressive disorder, but that depression is a concern that may warrant additional assessment.*

### **74%** Report Elevated Anxiety

*Using a self-report anxiety screen, nearly 2/3 reported elevated symptoms of anxiety. This does not necessarily mean that 74% have an anxiety disorder, but shows that anxiety is a common concern that may warrant additional assessment and/or treatment.*

## Many Experience Peer Victimization



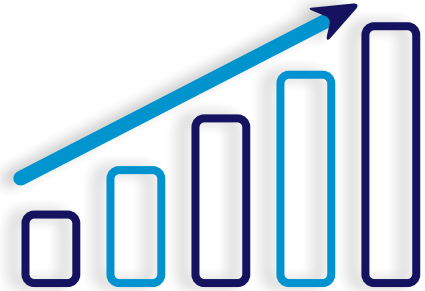

### Overt

*More than half report that peers sometimes or often yell at them or call them mean names. Nearly half report sometimes or often being threatened by peers if they don't do something for the peer. Approximately 1/4 report often being hit and pushed by peers.*

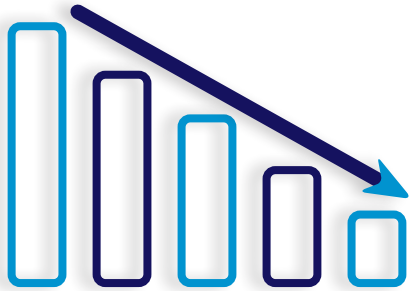
### Relational

*More than half report that peers sometimes or often exclude them on purpose. One quarter to one third report that peers gossip or lie about them to peers, manipulate them to do things, or retaliate against them.*

# Peer Relationships are Associated with Symptoms of Depression and Anxiety



*People with mDs who reported experiencing more relational and overt peer victimization also reported more anxiety and depression symptoms.*

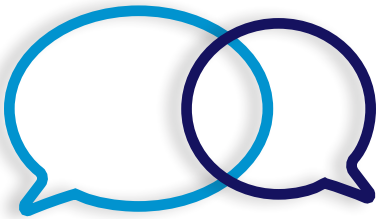


*But, those who reported experiencing kindness from peers reported experiencing LESS anxiety and depression.*

## Prevention and Intervention

This study is a first step in understanding the mental health needs of people with mosaic Down syndrome and more research is needed. But there is every reason to believe that people with mDs would benefit from prevention and intervention strategies as much as people without mDs. Here are some strategies that might help if you or your child are experiencing symptoms of depression, anxiety, or peer relationship problems.

*Talk with a mental health professional. Cognitive-Behavioral Therapy and Dialectical Behavior Therapy have been shown to be effective for people with and without learning disabilities.*



*Talk with your child, or someone you trust, about what's been going on. Keep lines of communication and be accepting of emotions.*

*Get a physical. Check thyroid, sleep apnea, and other health conditions that can look like depression or anxiety.*

*Develop healthy peer relationships. Help your child find groups and activities that are supportive and accepting of all abilities. Help your child learn and develop healthy social skills through coaching, skills training, and plenty of opportunities for practice.*

*Stand up to bullies. It is no longer recommended to "just ignore" bullies. Most schools and workplaces have policies against bullying. Bullying people with disabilities is also against the law. See stopbullying.gov and Pacer's National Bullying Prevention Center for resources on responding to bullying.*


*Family Connect Day Outing, Research and Retreat Weekend. Nashville, TN, 2019*

# www.IMDSA.org

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