“Uniting Unique People in a Unique World.”

International Mosaic Down Syndrome Association

Information for Families & Professionals

www.IMDSA.org
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.

- **IMDSA website** ([www.imdsa.org](http://www.imdsa.org)) that serves as a first introduction to any families and professionals learning about mDs and IMDSA.

- **New Family Welcome Kit** gives new families information to help answer questions they may have about mDs and how to deal with a new diagnosis for their loved one.

- **Monthly Newsletter** communicates to IMDSA membership the latest activities of IMDSA and the Board of Directors, helpful tips for parents and personal stories of those whose lives have been touched by mDs.

- **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.

- **Biennial Research and Awareness Conference** that allows families to connect with each other and attend educational workshops to help them support and advocate for their loved one with mDs. This conference also allows professionals interested in mDs (or Down syndrome without mosaicism) to conduct research with attendees at the Conference.

IMDSA is more than just an organization. IMDSA is a family of caring individuals who are there to support families and professionals.
What is mosaic Down syndrome?

Medically, it is known as mosaicism for trisomy 21. Throughout this document it will be referred to as mosaic Down syndrome or mDs. It happens when an individual has 2 or more types of cells; usually a percentage of cells with an extra copy of chromosome number 21, as well as a percentage of cells that have the typical number (2 copies) of chromosome 21 (unaffected). This condition occurs as a result of chromosome sorting problems that arise prior to and/or after conception and is no fault of the parents.

According to research, 2-4% of the Down syndrome community have mDs. However, this number does not account for the individuals who never receive a diagnosis or those who are misdiagnosed with complete Down syndrome.

- Average age of diagnosis is 1-4 years old
- Not restricted to any race, culture or religion
- Physical characteristics may not be present
- IQ levels generally 10-30 points higher than those with complete 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
- The learning potential will vary between individuals, but some adults go to college, marry and have children
- Children may or may not have Down syndrome depending on if affected cells are in reproductive organs
- Research shows that siblings of those with Down syndrome/mDs are well adjusted and often take careers in medicine, law or education
- Translocation, another type of Down syndrome, accounts for about 4% of all cases. Just as with complete Down syndrome, mosaicism for translocation can also occur.
**Laboratory Logo**

**Chromosome Analysis**

**Name and address of Testing Laboratory**

**Patient Name:**
**Referring Physician:**
**Specimen #:**
**Client #:**
**DOB:**
**Date Collected:**
**Date Received:**
**SSN: ____-____-____**
**Lab ID #:**
**Hospital ID #:**
**Indications for Study:**
**Specimen Type: Peripheral Blood**
**Clinical features of Trisomy 21**

**Metaphases Counted:** 21
**Metaphases Analyzed:** 6
**Metaphases Karyotyped:** 2
**Number of Cultures:** 2
**Banding Technique:** GTG
**Banding Resolution:** 550
**Dept. Section:** B2

**RESULTS:** 47,XX,+21(15)/46,XX(8)

**Abnormal karyotype, female**

**INTERPRETATION:**
Cyto genetic analysis shows three copies of chromosome 21 (trisomy 21) in fifteen out of 21 metaphase cell examined. An additional 10 cells were scored for the presence of an extra copy of chromosome 21 and all 10 cells were trisomic. This is consistent with the clinical diagnosis of mosaic Down syndrome.

Due to the presence of mosaicism the phenotypic features may be variable. The most common manifestations include mental retardation, cardiac anomalies (40%), relatively small stature, neonatal hypotonia and characteristic facies. Social development is more advanced than intellectual development. Infants with heart defects have a high mortality risk, but otherwise life expectancy is close to normal (Jones, K.L., Smith's Recognizable Patterns of Human Malformation, 4th edition. Philadelphia: W.B. Saunders Co, 1988. Pp. 10-15).

An increased risk for recurrence exists for this or other numerical chromosome abnormalities.

No other chromosome abnormalities are observed. The standard cytogenetic methodology utilized in this analysis does not routinely detect small rearrangements and low level mosaicism, and cannot detect microdeletions.

**Genetic counseling is recommended for this family.**

**Signature of the Cytogeneticist Reviewing the Case and the Date of testing**
Cytogenetic Diagnosis

To diagnose mDs, a genetics laboratory must evaluate the individual’s chromosomes in many cells. At least 30 cells should be counted and many cytogenetic labs will score up 100 or more cells (depending on the technique used).

It is important to recognize that the proportion of trisomic cells varies 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 cell for diagnostic testing.

Explaining Test Results/Karyotypes to Parents

A sample test result is shown to the left. Our goal is to help you interpret these results.

1. The specimen type indicates that this particular test looked only at blood cells and not at cells of any other tissues (for example, skin cells). Because these cell types have different origins, they could have different percentages of cells with mDs. 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 occurs when cells are dividing. Since the DNA condenses at this stage, it is possible to count the number of chromosomes present.

Metaphases Counted: the technician counted the number of chromosomes present in the indicated number of cells, but did not evaluate them all on a band-by-band detailed manner.

Metaphases Analyzed: the technician looked at the chromosomes (band-by-band) to look for small amounts of extra or missing genetic information. In the example report shown, the technician looked at all of the bands on all of the chromosomes from 6 cells.

Metaphases karyotyped: a picture (karyogram) was prepared in which the chromosomes were aligned in an ordered arrangement (see #5).

3. 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 child (and not artifact from the lab growth process). In this case, two independent culture vessels were used. Results from two or more cultures are more reliable than results from one.
4. The banding techniques and resolution describe the methods used for these studies. For mDs or Ds, it is not necessary to have high resolution because an entire extra chromosome is present. Higher resolutions are necessary when looking at smaller amounts involving portions of a chromosome.

5. All of the organs (heart, liver, pancreas, etc.) in our body are made up of cells. Each cell contains the genetic information (an instruction manual) our body needs to grow and develop. The genetic information is packaged into chromosomes. There are 23 pairs of chromosomes, and each contains a different set of genetic instructions. Since our blood is also made up of cells, our chromosomes can be found there too. 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 types of chromosomes are the same in males and females. The sex chromosomes will differ between males and females. Females have two X chromosomes and males have one X and one Y chromosome. It is likely that your test results will look something like this: 46,XX.

This is just shorthand for the karyotype results. The number 46 indicates that there were 46 chromosomes present, and the XX indicates there were two X chromosomes observed, indicating that the person is female. If the individual was a boy, it would be designated as: 46,XY.

If your child has been diagnosed with Down syndrome, the karyotype could have an extra copy of chromosome 21. If the child were a girl, it would be written out as: 47,XX,+21 [indicating that this child has a total of 47 chromosomes, including two X chromosomes (which makes her a girl) 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:
In this case, the two different types of cells present in the individual studied 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 we would really like to know is **What does a mosaic karyotype mean for my child’s development?**

Unfortunately, we don’t currently have the answer to that question. That is why IMDSA strongly supports research so that we can better interpret such test results and predict what it means for management of individuals with mDs.

6. Many parents would like to know what the chances are of having another child with mDs. The “increased risk” listed on this results form is based on studies of children with Down syndrome. There are no large studies evaluating specifically the recurrence risk of mDs. However, the studies on Ds indicate that the chance of having a second child with Ds is about 1%. This is greater than the population risk of about 1 in 800.

7. A genetic counselor works with the genetics team 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. Often times genetic counselors are the ones who provide support group information and inform people of research studies that are ongoing for the condition of interest. They will also talk to you about recurrence risks for future pregnancies.
Developmental Milestones

As a parent of a baby or toddler with mDs you will be concerned for their developmental growth. It is important to remember that your baby or toddler will reach these important milestones. Sometimes your baby or toddler may need a little extra help to develop the skills they need to reach these important steps in development.

To help your baby or toddler reach these steps, a therapist may be needed. You can contact your physician or government offices to find out about the services available in your area.

It is important to not compare your baby’s development to others with or without mDs. This will only frustrate you and your baby. With you and your therapist’s help, he/she will reach these important milestones.

Not all babies are exactly alike! Each one will reach his or her milestones in his or her own time. This is just a guide for you to 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 important milestones! 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

<table>
<thead>
<tr>
<th>Milestone</th>
<th>Time</th>
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<tbody>
<tr>
<td>Roll Over</td>
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<tr>
<td>Sit w/o Support</td>
<td>8 - 9 months</td>
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<tr>
<td>Crawl</td>
<td>11 months</td>
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<tr>
<td>Drink from Cup Alone</td>
<td>17 months</td>
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<tr>
<td>Walk unassisted</td>
<td>20 - 21 months</td>
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<tr>
<td>Walk up stairs</td>
<td>24 - 25 months</td>
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Healthcare Guidelines: Birth—Five

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 copy of the 21 chromosome, they can develop many of the health problems associated with “full” 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 children of his/her age, but they may also complete some extra testing. Since children 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 yearly neck X-ray (typically starting a 3 years of age).

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

Please refer to the recommended Health Care Guidelines to help you know when your child may need these exams.
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<tr>
<th>Medical Issues</th>
<th>Birth</th>
<th>6 mos.</th>
<th>12 mos.</th>
<th>18 mos.</th>
<th>2 years</th>
<th>2.5 years</th>
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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
Motor Skills

Development of 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 causes differences in gait
- Low muscle tone and lack of strength commonly result in a rounded sitting posture

**Fine Motor Skills**

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

Referrals for Physical and Occupational therapy are often helpful.
Education

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 US, 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.

- Open communication about the IEP will be the key to success in the classroom. Communicate with the parents to let them know you are on their side and you plan to work as a team to help the student.

- 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.

- 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 paid or unpaid job experience with a job coach.
Counseling

• Congratulate the family on the birth.
• 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.
• Limit parental liability. Explain to the parents that nothing they did caused their child to have mDs.
• Help families to understand that the percentage of affected cells does not indicate what areas of health and development will be affected.
• Use “people first language”.
• Some families feel they are unable to deal with a child with 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 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, standard counseling for 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 Ds due to a structural chromosome change (such as a translocation) will have different risk that will be explained by the genetic counselor. Maternal age should always 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.
- There is a great deal of variability in the clinical findings of individuals having Ds, but this range is even broader in people having mDs.
- Characteristic facial traits include upward slanting eyes with epicanthal folds, 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/Ds have an increased risk for developing leukemia.
- Cognitive impairment in the mild to moderate range requires early intervention services soon after birth to address developmental and speech delays.
- Individuals with mDs may have higher cognitive functioning and their facial features may be more subtle when compared to people having Ds.
- 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, this correlation is imperfect.
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.com under Information> Information Packets> Down syndrome Medical Checklist.
Top Five 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 clients and patients.

Q. How is this going to affect my child’s development?
A. Because there is no way to know where the affected cells are located in the body, there is no way that we can predict how your child will be affected. With proper intervention your child has a great opportunity to 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 the child will develop these conditions. With proper medical intervention outlined in the Health Guidelines for Down syndrome your child can live a healthier life.

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 female with mosaic Down syndrome was noted to be 83 yrs old*. With current medical treatments available most individuals should live long fulfilling lives. * Brian Chicoine, MD & Dennis McGuire, PhD. 1997

Q. Will my child have children when he/she grows up?
A. This depends on where the affected cells are located in the body. If the affected cells are in the reproductive organs then the males may be sterile. However, we do know of many cases where individuals (both male and female) have had children and their children may or may not be affected by the extra chromosome.

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. Many people are often confused in the pronunciation of this chromosomal condition. It is not pronounced “Down’s” and there is no apostrophe “s”; it is simply “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, the professional. and it is very simple!
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.”