

WHAT IS  
WILLIAMS  
SYNDROME

# INTRODUCTION

My name is Gianna. I am 28 years old and live in New Jersey.

I was diagnosed with Williams Syndrome (WS) at the age of two and a half.



I graduated from high school in 2016 and went on to attend Centenary University, where I earned an associate's degree in Liberal Arts.



I then continued my education at Ramapo College of New Jersey, where I received a bachelor's degree in Music Performance.

# EVENTS & OPPORTUNITIES

Through Williams syndrome, I have met incredible people who's support, and connections have created opportunities that have shaped my journey.

I have had the opportunity to speak at Advertising Week in New York multiple times as an advocate for Creative Spirit.

Creative Sprite is an organization dedicated to creating employment opportunities for individuals with intellectual, developmental, and learning disabilities.

One unforgettable moment was meeting Sandra Swift Perrino, who contributed to the creation of the ADA and stood alongside President Bush at its signing.



Meeting Sandra Swift Perrino



ADA (Americans with Disabilities Act) being signed into law.

# WILLIAMS SYNDROME AWARENESS MONTH

I also had the opportunity to travel to Albany, New York, for the proclamation, declaring May as Williams syndrome Awareness month.



# MUSIC

Music is a central part of my life. I am a drummer and singer, and I have performed at Williams syndrome and Creative Spirit galas.

I had the opportunity to play the drums during a performance with Scarlet Burke and also with the band Hotel fiction. I sang with OAR and a recording artist Justin Edwards.



# Representation

Representation for people with disabilities matters everywhere!

If an actor without a disability plays someone with a disability it is sending a message, that we are not able to perform that role.

Growing up I have always wanted to be in the entertainment industry but I didn't have anyone to look up to. Roles of people with Williams syndrome were played by actors without disabilities. It made me frustrated and upset.



# BOARD OF TRUSTEES

I serve as a member of the Board of Trustees for the Williams Syndrome Association, where I am proud to contribute my perspective as a person with Williams syndrome and support the organization's mission. The WSA supports all families affected by Williams syndrome along with supporting and fundraising for much needed research.



# The Genetic Basis of Williams Syndrome

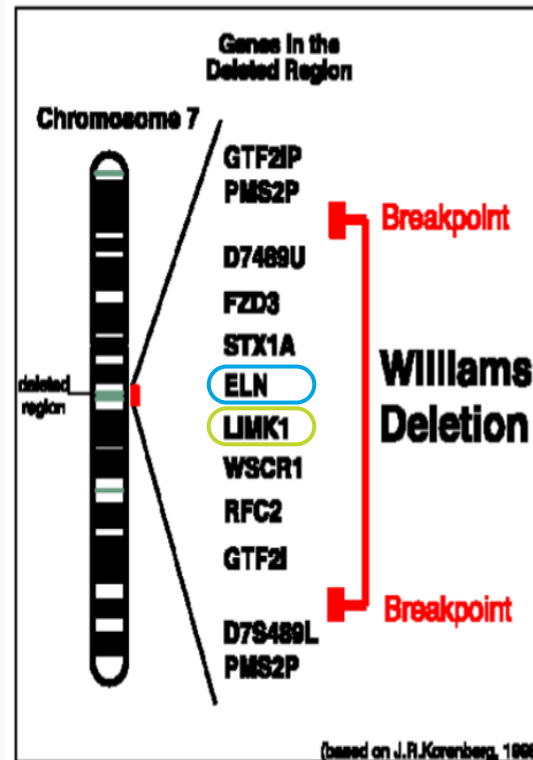
Deletion (missing piece) of chromosome 7q11.23

Approximately 26 - 28 genes live in that piece of deleted Material

WS is a “Contiguous Gene Deletion” syndrome (all the genes “line up” in one area)

Each gene has one or more jobs (function)

We now know the functions of some genes in the missing piece but not all the functions of every gene

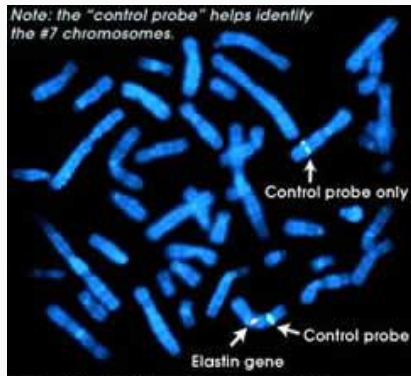


**ELN:** the gene for elastin, a protein that provides elasticity to organs and tissues.

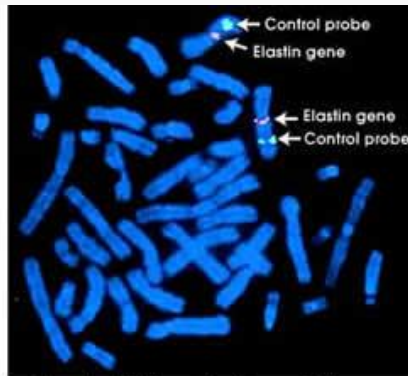
**LIMK1:** has been argued to be responsible for aspects of visuo-spatial cognition.

# DNA Tests to Diagnose

There are two DNA tests that can determine if a person has Williams syndrome. The FISH test and the Microarray test. These tests are looking to see if one of the 7th chromosomes is missing a patch of about 26- 28 genes.

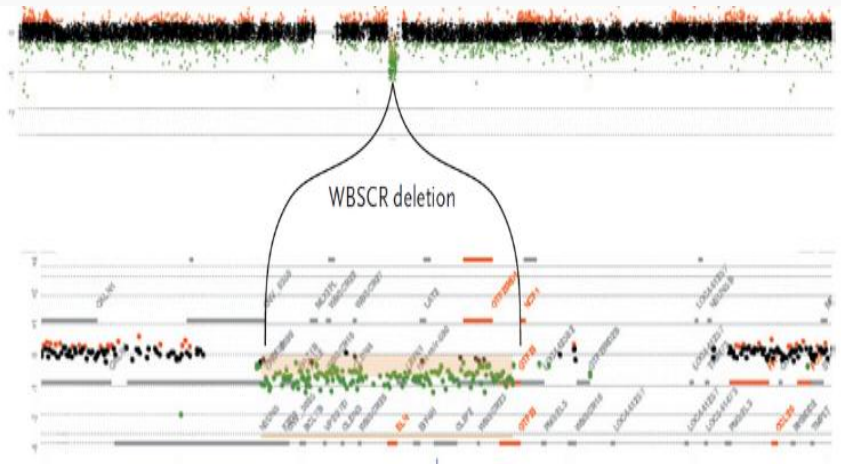


**Positive Williams Syndrome FISH assay (Chromosome 7)**  
The elastin gene is found on only one chromosome. The other copy carries an elastin gene deletion.



**Negative Williams Syndrome FISH assay (Chromosome 7)**  
The elastin gene is found on both chromosomes. This individual does not have Williams Syndrome.

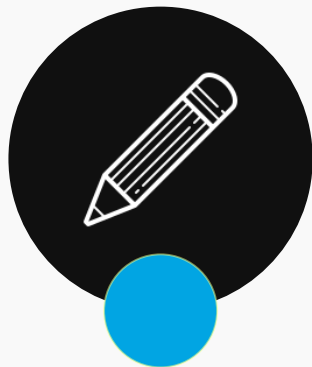
FISH Test



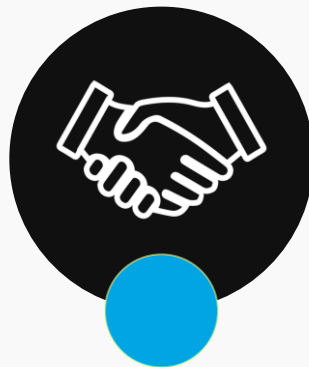
Microarray Test

# Characteristics of a Person with Williams Syndrome

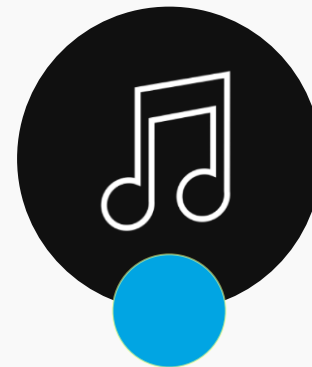
There are many characteristics of Williams syndrome. Some people have:



**Learning  
Differences**



**Very Friendly  
Personality**



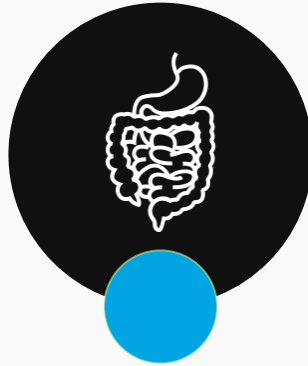
**Affinity for  
Music**

# Medical Complexities

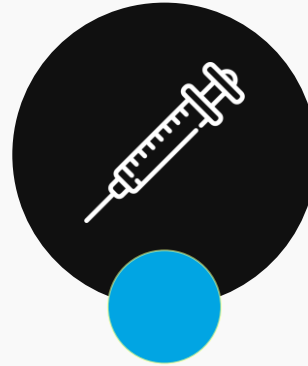
There are many medical Issues within the Williams syndrome community!



**Heart  
Defects**



**Gastrointestinal  
issue**



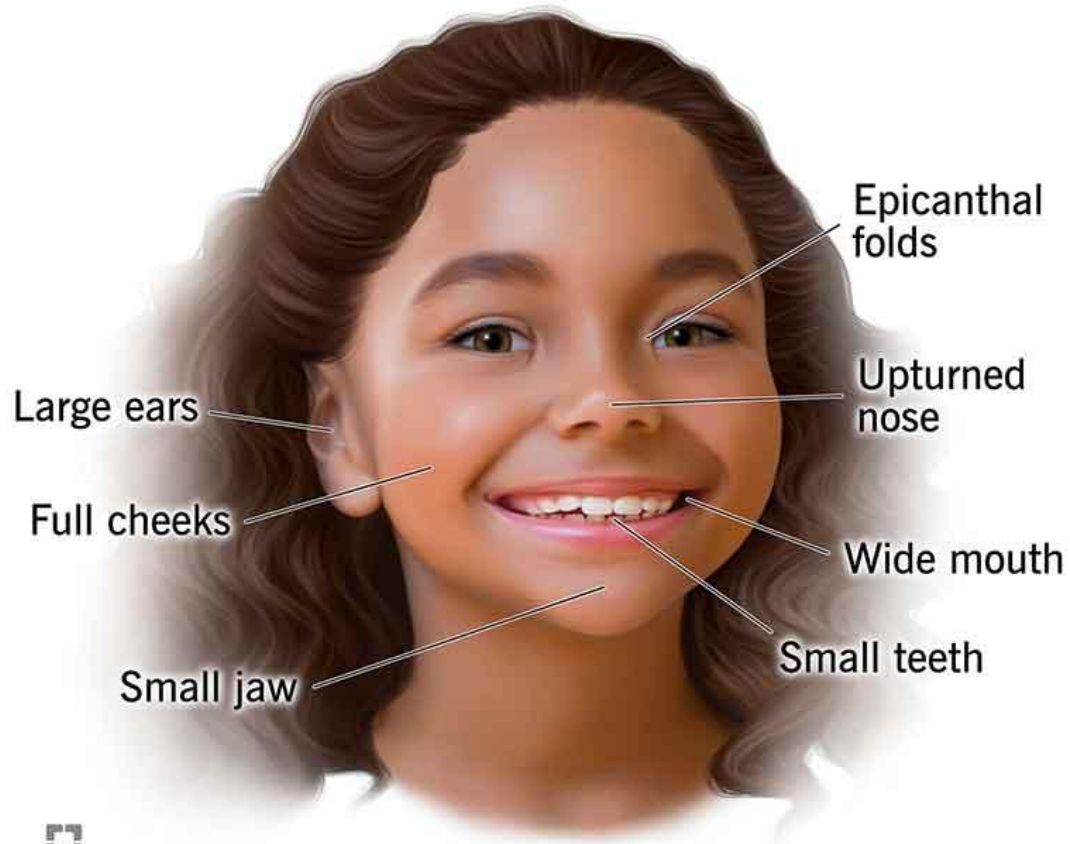
**Anesthesia**



**Sudden  
Death**

# FACIAL FEATURES

## Williams syndrome





## HEART DEFECTS



Cardiovascular abnormalities are common in individuals with Williams syndrome, affecting about 75% of those diagnosed. These conditions can vary greatly in both presentation and severity, ranging from mild heart murmurs to more complex structural defects that may require repeated intervention, including open-heart surgery. The most common cardiovascular findings include narrowing of the aorta (aortic stenosis), narrowing of the pulmonary arteries (pulmonary stenosis), and various abnormalities affecting the heart valves. These issues impact the blood vessels and cardiac structure.



## OPEN HEART SURGERY

Many of my friends with Williams syndrome have undergone open-heart surgery by the age of five. I also met a mom through Williams syndrome whose daughter Emma had already experienced three open-heart surgeries by the time she was four. She was later told that Emma would need another surgery, but that the risks were extremely high and she might not survive. Her doctors recommended waiting as long as possible to allow her body to grow stronger before attempting another procedure. For now, she is still waiting and I continue to keep her and her family in my thoughts and prayers.





## GASTROINTESTINAL ISSUES

Most individuals with Williams syndrome experience gastrointestinal (GI) issues at some point, with abnormal bowel patterns—particularly constipation—and associated complications such as rectal prolapse and hemorrhoids being especially common. Episodes of abdominal pain are also frequently reported. Research indicates that GI symptoms can represent a significant clinical concern in this population. Commonly observed conditions include gastroesophageal reflux, abdominal pain, constipation, diarrhea, irritable bowel syndrome, and various forms of GI dysmotility. Less common but clinically important issues include early-onset diverticulitis, celiac disease, and gluten intolerance.



# ANESTHESIA



Individuals with Williams syndrome are known to have an increased risk of anesthesia-related complications, and significant adverse events have been documented when anesthesia is not managed with appropriate vigilance. Clinicians are therefore advised to conduct a thorough review of the relevant medical literature before proceeding with any anesthetic intervention in a child with Williams syndrome. A comprehensive pre-anesthetic evaluation is essential, and maintaining adequate hydration until two hours prior to the procedure is recommended.



## SUDDEN DEATH

There have been numerous instances of individuals with Williams syndrome passing away unexpectedly. I have personally lost several friends in this way, which has been deeply distressing. In many cases, no definitive cause is identified. Although these losses are often presumed to be cardiac-related, many of the individuals were under regular cardiology care and showed no indications of a severe underlying issue.



DAREN

Two years ago, I had the privilege of meeting an extraordinary eight-year-old boy named Daren, who had Williams syndrome. We were introduced at a Williams syndrome convention in Arizona during a gathering for first-time attendees. As a member of the Board of Trustees, I was assigned to Daren's family as their mentor, and I quickly came to admire his joyful spirit and the warmth of his family.

A few months later, the Williams syndrome community received heartbreaking news: Daren had unexpectedly passed away while playing at home with his cousin. It was one of the most devastating and frightening events I had ever had, and it deeply affected everyone who had been touched by his light.

## RESEARCH GROUPS



I have many additional stories like the one I shared, each underscoring the urgent need for expanded research. Williams syndrome is classified as an orphan disease, which means it receives limited federal support and lacks the level of scientific investment required to address its complex medical challenges. As a result, families and caregivers are often left to shoulder the responsibility of funding and advancing research themselves.

One such parent is Camile Fortunato, who founded the AFG organization with the mission of driving progress in this area. Each year, AFG raises critical funds to support research led by Dr. Levin at the University of Pennsylvania, focused on understanding the causes behind the unexpected deaths occurring within the Williams syndrome community. Their work represents a vital effort to uncover answers, improve outcomes, and ultimately save lives.

# LEARNING DIFFERENCES

Educators face unique challenges teaching children with Williams syndrome, but with knowledge of their learning style, students with Williams syndrome can experience success in the classroom.

# STRENGTHS

1

**Sociable  
Nature**

2

**Expressive  
Vocabulary**

3

**Long-Term Memory  
for Information**

4

**Short-term  
& Long-term  
Auditory Memory**

5

**Hyperacusis**  
(helps with phonics but can  
make the child worried  
about loud noises)

# AREAS FOR CONCERN

1

**Attention difficulties**  
often distractible  
& impulsive but not  
necessarily hyperactive

2

**Visual-spatial  
& visual-motor  
integration  
difficulties**  
This makes writing  
difficult

3

**Visual Memory  
Deficits**

4

**Difficulty with  
Abstract Concepts  
& Reasoning**  
This makes math  
concepts difficult  
to grasp

5

**Perseveration on  
“Favorite” Topics**

6

**Difficulty  
Processing  
Nonverbal  
Information**

# Williams Syndrome Personality (considered the ‘opposite of Autism’)

People with Autism Spectrum Disorder (ASD) often experience differences in social communication and sensory processing. This can make social interaction more complex and tiring for them. Some individuals prefer less social contact. In contrast to individuals with Autism, Individuals with Williams syndrome are highly (overly) sociable and empathetic, often showing a strong drive to engage with others, even strangers. There is a major concern within the Williams syndrome community that these children will go with anyone so ‘stranger danger’ is taught at a very young age and continues through-out their lives. This highly sociable personality and strong desire for interpersonal connection, makes meaningful opportunities for social interaction especially important to them throughout their lives. At the same time, many individuals with Williams syndrome have difficulty interpreting subtle social cues, which can make it challenging to build and sustain long-term relationships. As they transition into adulthood and move beyond the structured environments of school and family routines, they may face increasing social isolation. This isolation can, in turn, heighten the risk of anxiety and depression, underscoring the importance of ongoing social support and inclusive community engagement.

# IMPORTANCE OF MUSIC



Music is very important to people with Williams syndrome. For my capstone project in college, I did a presentation about 'Williams Syndrome, Music, and the Brain'. The next few slides are from my capstone project. It talks about why music is so important and why music therapy is very helpful to a WS child.





THE NORTH POLE

SANTA'S WORKSHOP

NEW YORK CITY

HMC STUDIOS

IT'S ALL FUN AND GAMES





Why do musicians create music? Is it the need to express themselves? To bring out emotion in others?

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Many musicians create and perform music for the simple joy of doing so. If this musical expression can help others, then it only makes sense that we use it as a type of therapy.

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Music is known to affect a person's emotions in many ways. Some people are more affected emotionally than others. If a person has an emotional connection to music, they are more likely to benefit from a music therapy session.

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Many people with a disability called William's syndrome have a greater emotional connection to music. This could be due to the difference in structure of their brain.

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Is music therapy beneficial for people with Williams syndrome and does the structure of the brain have anything to do with it?



# HOW THE BRAIN IS IMPACTED BY MUSIC

## How the brain interprets music

Listening to music involves more than just ears

**Auditory cortex**  
Where sounds and tones are perceived and analyzed

**Nucleus accumbens and amygdala**  
Where emotions related to music are processed

**Hippocampus**  
Part of interpreting music memories and experiences

**Cerebellum**  
Part of emotional and physical reactions while experiencing music

SOURCE: Music for Young Children

Research suggests that the way emotions are generated by music is due to different structures of the brain.



## STRUCTURE OF A WILLIAMS SYNDROME BRAIN

There are many MRI research studies about the structure of the brain of people with Williams syndrome. One of the experts in this field is named Ursula Bellugi, who I have had the honor of talking to in the past.

Dr. Bellugi worked at the SALK institute in LA and is an expert in this field of research. In one of her studies, it says that the Williams syndrome brain is different than a typical brain. One of the ways it is different is due to the size and structure of the amygdala section of the brain, where emotions that are related to music are thought to be processed.

According to the article Neural Correlates of Auditory Perception in Williams Syndrome: An MRI Study, when music was played during the MRI, there was "greater activation in the right amygdala for the WS group compared to the control group, pointing to a potential dissociation: control subjects show greater activation in the temporal lobes, and WS participants (who have a known affinity for music) have greater activation in the amygdala" (Neural Correlates of Auditory Perception in Williams Syndrome: An .. n.d.).



## LEARNING DIFFERENCES

I created a survey to get more insight on a person with Williams syndrome's emotional response to music. I asked the Williams Syndrome Association to put my survey on all their private member Facebook pages. I limited the people's responses to those who specifically said they had music therapy. Since Williams's syndrome is so rare, I was happy to get 50 responses on my survey. And I believe this is a good pool of people that responded.

One of the questions I asked was "has your child ever had an unusual reaction to music, and if so, what emotion did they portray?"

I created a chart that shows all responses that I received, with happiness being the most at 80%, followed by 65% for excitedness, and sadness came in a little lower with 50%. The chart also shows that over 92% of people with Williams syndrome had an emotional connection to music.



## A PARENTS STORY

The responses I received from the parents about their child's emotional response to music were partially what I expected with how many people responded with happiness and excitement. What was interesting is that over half of the people's responses were sad. I do not feel that this would be a typical response for so many people to have, since most of the responses were about children under the age of five.

I decided to send out additional questions to the parents that responded with sadness as an emotional response to music in the survey. I asked the parents to describe what happened during the sad emotional response.

All the parent's descriptions were similar. One parent described their child's unique response to music as follows.

"The first time it happened was when my daughter was about 18 months old. When we were at a birthday party, everyone was happy and excited, and we started singing Happy Birthday. I saw the look on my daughter's face change, big tears welled up in her eyes, and she began sobbing uncontrollably. I did not understand what was wrong. It happened again at another person's party.

When I got home that evening, I started singing Happy Birthday to her.

There were huge tears and she started sobbing. When I stopped singing, she looked at me and said 'Again'"



## CONCLUSION

According to my research, **people with Williams syndrome have a true emotional connection to music** and children that are as young as 18 months old have been known to have deep emotional responses to music

As I mentioned above, **the amygdala and hippocampus are parts of the brain known to be related to the processing of music.** Through research on brain structures, using functional MRI's, the amygdala and hippocampus part of the brain within a person with Williams syndrome responds differently to music than a typical person.

My research shows that people with Williams syndrome have a stronger response when music is played. After reviewing the results from the survey, I sent out to the parents/caregivers, and reviewing the research about the brain of a person with Williams syndrome, music therapy can be used to help people with Williams syndrome in many different areas of their life

## WRAP UP

As you can tell from this information I shared with you, Not many people know or understand how rare William's syndrome is and how much we need funds for more research. It is so important because of how complex our medical issues are and because of all these people passing away unexpectedly!

I want to thank you all from the bottom of my heart for coming to my webinar today!! Down on the next slide I have linked some websites you guys should check out for more information and if you would like to donate to the Williams Syndrome Association and to the Anthony Filippazzo Grant for Williams Syndrome Research, That would be amazing!

Have a great day!!

# KEY INFORMATION

Website for more information about Williams syndrome

<https://www.williams-syndrome.org>

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Website for more information about research for Williams syndrome

<https://wsresearch.org/>

# MY INFORMATION

Gianna Morello

GMorello@ArcNJ.org

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## **Facebook**

Gianna Morello (Gigi)

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## **Instagram**

@GigisJourney26