

## Medeiros Family

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Juan and Cecelia Medeiros brought their son, Manuel, to the genetics counseling clinic because he was beginning to show symptoms similar to those of his cousin David who has Duchenne's Muscular Dystrophy (DMD). They are very concerned because David, age 18, is now confined to a wheel chair and he is not expected to live much past his twentieth birthday.

Duchenne's Muscular Dystrophy is a condition involving the gene that codes for the protein *dystrophin*, a large protein that plays a role in the contraction of muscle cells. Without the full function of dystrophin, persons with the condition begin to experience muscle degeneration before three years of age, are often confined to a wheelchair by age 12 and frequently die by age 20. Muscles involved in breathing and heartbeat are also affected so heart and respiratory failure contribute to DMD being a lethal condition. Fortunately DMD is extremely rare, occurring in less than 7 of every 100,000 births in the U.S..

Because the muscles work with the skeleton to move and support the body, individuals with DMD can also have skeletal problems such as scoliosis. Mild cognitive disability can occur, although it is not typical, as dystrophin is also produced in the brain.

Name Manuel Medeiros Sex Male  
Age 2 years Birth weight 7lbs 9oz Birth length 19 inches

Were there any complications or problems at the time of birth? If yes, please explain.

No

Complete the following, including ages and dates where known.

SURGERIES OR HOSPITALIZATIONS:

ILLNESSES, CHRONIC INFECTIONS AND OTHER HEALTH PROBLEMS

SPECIAL TREATMENT, THERAPY, PRESCRIPTIONS

Child's current height 36 in weight 31 pounds

Describe behavioral/emotional concerns:

What is it about your child that causes concerns?

He is beginning to show some of the same symptoms as his cousin David did when he was Manuel's age.

When were these concerns or problems first noted?

Within the last few months

What have you been told about the child's problems?

Our pediatrician recommended that we see a genetic counselor because he thought Manuel might have some type of muscular dystrophy.

What questions do you want to get answers for at this time?

Manuel's cousin David is confined to a wheelchair and is not expected to live much longer. We are very concerned about our son's future.

Name of person completing this form Cecelia Medeiros relationship mother

Clinical Genetics Center  
Family History

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Subject's Name: Manuel Medeiros Age: 2 years.

Brothers/Sisters

(Please include all brothers/sisters, half-brothers/sisters, living and deceased, including pregnancy losses and stillbirths.)

| Name and age        | Sex | Health/medical problems; birth problems or defects; physical, mental, or developmental concerns |
|---------------------|-----|---|
| Rita<br>5 years old | F   | none  |
|                     |     |   |
|                     |     |   |
|                     |     |   |

Are any of the parents or grandparents of the above children related to each other? If so, please explain.

No

Is there anything else in the personal, medical, or family histories which you want us to know about?

Nothing unusual

Father's name: Juan Medeiros Age: 33 years  
Height: 5' 10" Occupation: professor

Health/medical problems, surgeries, hospitalizations, medical testing/treatments;  
physical, mental concerns; congenital problems or unusual features.

Juan is nearsighted. He broke his leg when he was 10 years old.

Father's brothers and sisters. Living and deceased.

| Name and age        | Sex | Health concerns | His/Her children Name, age, sex | Health concerns |
|---------------------|-----|-----------------|---------------------------------|-----------------|
| Antonio<br>38 years | M   | none            |                                 |                 |
| Elisa<br>36 years   | F   | none            |                                 |                 |
|                     |     |                 |                                 |                 |

Father's mother's name: Teresa Medeiros Age: 63  
years

If deceased, cause of death and age: \_\_\_\_\_

Father's father's name: Diego Medeiros Age: 63 years

If deceased, cause of death and age: \_\_\_\_\_

Are there any other family members not listed here with birth defects, handicapping conditions, mental retardation or other concerns? If so, please explain.

Clinical Genetics Center  
Mother's Family

Mother's name: Cecilia Medeiros Age: 33 years.

Height: 5' 3" Occupation: journalist

Health/medical problems, surgeries, hospitalizations, medical testing/treatments;  
physical, mental concerns; congenital problems or unusual features.

I had an emergency appendectomy when I was in high school.

Mother's brothers and sisters. Living and deceased.

| Name and birthdate | Sex | Health concerns | His/Her children Name, age, sex | Health concerns                    |
|--------------------|-----|-----------------|---------------------------------|------------------------------------|
| Jorge<br>36 years  | M   | none            |                                 |                                    |
| Linda<br>39 years  | F   | none            | David<br>18 years               | David has<br>muscular<br>dystrophy |
|                    |     |                 |                                 |                                    |
|                    |     |                 |                                 |                                    |

Mother's mother's name Maria Rodriguez Age: 59 years

If deceased, cause of death and age: \_\_\_\_\_

Mother's father's name: Luis Rodriguez Age: 61 years

If deceased, cause of death and age: \_\_\_\_\_

Are there any other family members not listed here with birth defects, handicapping conditions, mental retardation or other concerns? If so, please explain.

Maria had a brother, Gustavo, who died when he was 18 years old. For the last 8 years of his life he was in a wheelchair.