

Marcus Family

Joy and Aaron Marcus have come to the genetics center because they are concerned about their newborn daughter, Janice. Both Joy and Aaron are achondroplastic dwarfs. They met at a meeting of a group called the Little People of America which is also where Joy's parents met. Joy's parents both have achondroplasia.

Individuals with achondroplasia are dwarfed in stature mainly due to a shortening of the long bones of the arms and legs. They have a long, narrow body trunk. Their heads are large, with prominent foreheads. Most are of normal intelligence and have a normal life expectancy. The skeletal growth features of achondroplasia are caused by problems with certain protein molecules on the surface of their cells that are supposed to act as receptors for a molecule called fibroblast growth factor 3 (FGFR3). Our bodies produce several different kinds of fibroblast growth factors. FGFR3 seems to play a role in the formation of certain types of bones, such as those in the arms and legs.

Patient Name Janice Marcus Sex Female

Age 1 month Birth weight 4lbs 5 oz Birth Length 16 in

Name of person completing this form: Joy Marcus Relationship to patient mother

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

Low birth weight

Complete the following, including ages and dates where known:

SURGERIES OR HOSPITALIZATIONS:

Currently hospitalized because of breathing problems

ILLNESSES, CHRONIC INFECTIONS AND OTHER HEALTH PROBLEMS

SPECIAL TREATMENT, THERAPY, PRESCRIPTIONS

Child's current height 16 in Child's current weight 4lbs 4 oz

Describe behavioral/emotional concerns:

What is it about your child that causes concerns?

She is in critical condition and seems to be more severely affected than her brother or sister

When were these concerns or problems first noted?

At birth

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

That she also has a severe dwarfing condition

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

Why is Janice more severely affected than the rest of the family?

Patient's Name Janice Marcus Age: 1 month

Patient's 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
Eric 3 years old	M	Diagnosed with achondroplasia
Kyle 6 years old	M	Normal Height
Anna 8 years old	F	Diagnosed with achondroplasia

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?

Not that we are aware of.

Clinical Genetics Center
FAMILY HISTORY

Patient's Father's Name Aaron Marcus Age: 39 years

Height: 4'10" Occupation: writer

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

Achondroplasia

Father's brothers and sisters (living and deceased):

Name and age	Sex	Health Concerns		His/Her children Name, age, sex	Health concerns
Ruth 46 years	F	none		Ken, 16 years, male Amy, 13 years, female	none
Natalie 34 years	F	achondroplasia		Jon, 14 years, male Melanie, 7 years, female	none achondroplasia
Tim 29 years	M	none			

Father's mother's name: Barbara Marcus Age: 69 years

If deceased, cause of death and age:

Health concerns: **Barbara has achondroplasia.**

Father's father's name: Tom Marcus Age: 68 years

If deceased, cause of death and age:

Health concerns: **None. Tom is of normal height**

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

No.

Clinical Genetics Center
FAMILY HISTORY

Patient's Mother's Name Joy Kirkwood Marcus Age: 38 years

Height: 3'9" Occupation: computer programmer

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

Achondroplasia

C-section delivery for all four children

Mother's brothers and sisters (living and deceased):

Name and age	Sex	Health Concerns		His/Her children Name, age, sex	Health concerns
Carl 34 years	M	none		Bruce, 8 years, male Robert, 5 years, male Ann, 2 years, female	none
Paul	M	Paul was severely dwarfed and died shortly after birth.			

Mother's mother's name: Judith Kirkwood Age: 60 years

If deceased, cause of death and age:

Health concerns: **Judith has achondroplasia:**

Mother's father's name: Daniel Kirkwood Age: 59 years

If deceased, cause of death and age:

Health concerns: **Daniel has achondroplasia.**

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

No