

Summers Family

The Summers family recently had their second child, Jane, who was diagnosed with PKU at the time of her birth. The parents, Joe and Angie, visited a genetics counselor after Jane's birth to learn more about their daughter's disease. The following pages are the forms that Joe and Angie filled out in preparation for their visit to the counseling center.

PKU (phenylketonuria) is a metabolic disease that affects the way the body processes phenylalanine, one of the amino acid building blocks that make up proteins in all organisms. People with PKU have a deficiency of *phenylalanine hydroxylase*, an enzyme (protein) necessary for normal processing of phenylalanine. Because they lack this enzyme, phenylalanine builds up in their bloodstream, causing brain damage and, subsequently, cognitive disability.

Children born with PKU appear normal for the first few months. If left untreated, they begin to lose interest in their surroundings by 3-5 months and by the time they reach 1 year of age they are cognitively delayed. They are often irritable, restless, and destructive. They may have a "musty" odor about them and often have dry skin or rashes. They also are unusually pale compared to others in their family.

Since the 1960s, children in all 50 states are routinely screened for PKU, within the first few days of birth, by a blood test that detects levels of phenylalanine. If the levels are above normal, the infant is placed on a special diet that is low in proteins containing phenylalanine. Foods that they avoid include breast milk, regular baby formula, cow's milk, cheese, meat, fish, and eggs. Since protein is essential for normal growth, they are given a special formula that is high in protein and essential nutrients, but has little or no phenylalanine. Individuals with PKU should remain on a restricted diet throughout childhood, perhaps for life.

Name Jane Summers Sex Female
Age 1 month Birth weight 6lbs 8oz Birth length 21 in

Were there any complications or problems at the time of birth? If yes, please explain.
Jane was diagnosed with PKU from a blood test done when she was a newborn

Complete the following, including ages and dates where known.

SURGERIES OR HOSPITALIZATIONS:

ILLNESSES, CHRONIC INFECTIONS AND OTHER HEALTH PROBLEMS

SPECIAL TREATMENT, THERAPY, PRESCRIPTIONS

We must keep her on a diet low in phenylalanine

Child's current height 21 in weight 6lbs 14oz

Describe behavioral/emotional concerns:

none

What is it about your child that causes concerns?

We worry that her brain development will be affected if we don't carefully follow the diet. She is also has a very light complexion so we worry that she will have trouble with overexposure to the sun.

When were these concerns or problems first noted?

Since birth

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

The special diet is important. If we do not follow it she could become cognitively delayed.

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

Will she need to be on the diet for her whole life?

Name of person completing this form Joe and Angie Summers relationship parents

Subject's Name: Jane Summers Age: 1 month.

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
Josh 3yrs	M	Allergies to pollen and dust

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

Father's name: Joe Summers Age: 32 yrs
Height: 6' Occupation: restaurant manager

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

nearsighted

Father's brothers and sisters. Living and deceased.

Name and age	Sex	Health concerns	His/Her children Name, age, sex	Health concerns
John 35 yrs	M	none	Stacy, 10 yrs Megan, 7 yrs Matt, 5 yrs	
Jill 29 yrs	F	asthma	Erin, 1 yr,	

Father's mother's name: Betty Summers Age: 56 yrs

If deceased, cause of death and age: _____

Father's father's name: William Summers Age: 57 yrs.

If deceased, cause of death and age: _____

Betty has Krohn's disease. William is nearsighted and has high blood pressure

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

Betty's aunt (her mother's sister Evelyn) was cognitively delayed but the cause was not known.

Clinical Genetics Center
Mother's Family

Mother's name: Angie Summers Age: 33 yrs.

Height: 5' 5" Occupation: attorney

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

Appendectomy at age 5.

Mother's brothers and sisters. Living and deceased.

Name and birthdate	Sex	Health concerns	His/Her children Name, age, sex	Health concerns
Tom 27 yrs	M	Has PKU		

Mother's mother's name Margaret Samuelson Age: 55 yrs

If deceased, cause of death and age: _____

Mother's father's name: Donald Samuelson Age: 55 yrs

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.

None known.