

J37 Newborn Screening

September 2009

Intro: Few words can capture the grief a family feels when they lose a child. The family you're about to meet said good-bye to two-year-old Makayla when she died from a rare genetic disorder. Many genetic disorders can be caught early if babies are screened for them soon after birth. But newborn screening did not include the disorder Makayla had. And while there's no known cure for that condition, her family is passionate about promoting newborn screening in hopes of saving lives.

Video

Total running time 1:50

Home video

CG :12 to :16

Stacy Pike

Makayla's mom

Audio

LITTLE ALLISON LUKEN NEVER
GOT TO MEET HER BIG SISTER
MAKAYLA.

"WHAT YA DOING, MAKAYLA?"
YOU SEE MAKAYLA WAS BORN
WITH A GENETIC DISORDER THAT
CAUSED HER CENTRAL NERVOUS
SYSTEM TO FAIL.

"SHE LOST THE ABILITY TO SMILE,
LOST THE ABILITY TO COO."

MAKAYLA'S MOM STACY PIKE
WATCHED HER BABY DAUGHTER
SLOWLY LOSE THE ABILITY TO
EAT, WALK OR MOVE AT ALL.

“SHE ESSENTIALLY WAS LIKE A WET NOODLE. SHE HAD NO MUSCLE TONE WHATSOEVER.” BUT IF YOU LOOKED DEEP INTO MAKAYLA’S EYES, GRANDMA SUSAN ROSENAU SAYS, YOU’D SEE THE WONDERFUL LITTLE GIRL WITHIN.

CG :36 to :46
Susan Rosenau
Makayla’s grandmother

“EVEN THE DAY SHE PASSED AWAY I DIDN’T THINK WAS AS HARD AS THE DAY WE GOT THAT DIAGNOSIS, KNOWING THAT SHE WOULD NOT BE WITH US FOREVER.”

SUSAN AND HER HUSBAND PAUL ARE TEAMING UP WITH DOCTORS AT MAYO CLINIC TO ENCOURAGE THE EXPANSION OF NEWBORN SCREENING TO DETECT MORE TREATABLE GENETIC DISORDERS.

CG :54 to :58
Piero Rinaldo, M.D., Ph.D. (23:50)
Mayo Clinic

“NEWBORN SCREENING SAVES LIVES. IT’S A FACT.”

DR. PIERO RINALDO SAYS

CG 1:06 to 1:16
Dietrich Matern, M.D.
Mayo Clinic

SCREENING IS SIMPLE TO DO.

BABIES GET A HEEL STICK TO
RETRIEVE THREE, FOUR OR FIVE
DROPS OF BLOOD.

“THESE BLOOD SPOTS ARRIVE IN
THE SCREENING LAB AFTER THEY
WERE COLLECTED ON THE
SECOND DAY OF LIFE. THE BLOOD
IS SPOTTED ON THIS FILTER
PAPER.”

DR. DIETRICH MATERN SAYS
MINNESOTA SCREENS FOR 54
GENETIC DISORDERS, THE ENTIRE
PANEL RECOMMENDED BY THE
AMERICAN COLLEGE OF MEDICAL
GENETICS. SOME OF THESE
DISEASES CAN BE TREATED IF
CAUGHT EARLY. BUT, STILL BABIES
DIE BECAUSE THERE ARE
DISEASES THAT ARE NOT
SCREENED. MAKAYLA’S FAMILY IS
WORKING TO CHANGE THAT.

Standup

THE ROSENAU’S HOPE THAT THE

CG
Vivien Williams
Reporting

MEMORY OF MAKAYLA'S SHORT
LIFE WILL HELP SAVE THE LIVES
OF OTHERS.

"IF WE CAN SAVE ANOTHER CHILD
FROM THIS, ANOTHER FAMILY
FROM GOING THROUGH THIS, IT'S
MORE THAN WORTH IT."

FOR MEDICAL EDGE, I'M VIVIEN
WILLIAMS

Anchor tag:

You might wonder if newborn screening is at all dangerous for the baby. Dr. Rinaldo says the answer is no. For more information about newborn screening, visit our website at...**[STATIONS: Per the licensing agreement, please provide a link from your station's website to <http://www.mayoclinic.org> or voice tag "mayoclinic.org" for more information.]**