

Public Comment to the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

Jill Fisch, parent

June 8, 2004

My name is Jill Fisch. I live in Scarsdale, New York with my three children Zach (12), Sara (8) and Matthew (3 1/2), and my husband Peter. I am the New York State Monitor for Save Babies Through Screening. I also am a parent representative on the Newborn Screening Task Force and the President of Matthew's Mission. I also have recently started working with Hunter's Hope regarding Newborn Screening issues. My son Matthew suffers from Short-Chain Acyl-CoA Dehydrogenase Deficiency (SCAD). SCAD is a disorder in which the cellular enzyme responsible for processing short chain fatty acids is missing from the cells or working at a diminished capacity. This disorder can cause Failure To Thrive, developmental delays, hypotonia or even death. We started Matthew's Mission to promote newborn-screening awareness as well as raise money for scad research. I became very involved with newborn-screening when I realized that after spending 2 years trying to get Matthew diagnosed, this was something that could have been screened for at birth. Matthew now has a feeding tube, significant hypotonia, and various other issues. After finding out he was carnitine deficient, he was started on Carnitor and gained a tremendous amount of weight. If we had known from birth, and he was started on the regimen he is on today, it is quite possible he could have had a different outcome. We will never know what Matthew's full potential could have been because he suffered do many setbacks while we were looking for a diagnosis. Finding out about Matthew caused us to find out that I also have scad. Matthew probably saved my life. Our rights as parents were taken away as we were not informed of supplemental testing. New York has the equipment to test for 60 disorders, but currently only screens for 11. One answer to this problem would be for the states to use resources in the private sector to provide the supplemental screening. Most parents do not realize that screening occurs or that options for more comprehensive screening are currently available through private labs. Unfortunately, we were one of those families. New Jersey will soon enact legislation mandating that parents be informed of additional tests available but not offered by their state program. This should be true of every state. Parental Notification of Supplemental Screening must be made mandatory. Babies are being born everyday and many are suffering adverse consequences from lack of screening. Comprehensive screening ensures that newborns are getting the best chance of starting a healthy life. I wish that Matthew had that chance. Congress should require states to inform parents in writing regarding outside screening through private labs. States that do not screen for all disorders should contract with an outside source to provide the comprehensive screening until the states are capable of doing the testing themselves. Mississippi screens for all disorders through a private lab with fantastic results. Don't all of our babies deserve the same chance? We feel our rights as parents were taken away since we were never informed of supplemental screening. This is something that can be changed and is being changed in different states as we speak. We want to see every family give their baby the healthy start that it deserves. That is the goal of Matthew's Mission. Thank you for giving me the opportunity to share. Jill Fisch

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