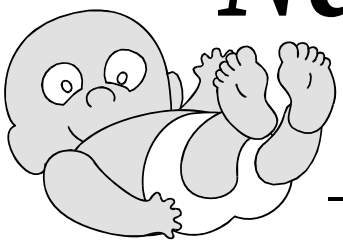

Newborn Screening News

Newborn Metabolic Screening • Newborn Hearing Screening

Colorado Department of Public Health and Environment • Health Care Program for Children with Special Needs (HCP)

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Health Care Program for children with Special Needs

Colorado Department of Public Health and Environment

ON YOUR MARK... GET SET....

Colorado to Begin Expanded Newborn Screening July 1, 2006

On July 1, 2006, the scope of the Colorado newborn screening program will increase from the current seven metabolic conditions plus hearing to a total of 29 conditions – 28 metabolic diseases plus hearing.

This panel of conditions, which has come to be known as “expanded newborn screening,” is the panel of screening conditions recommended by the American College of Medical Genetics (ACMG) and the March of Dimes. The ACMG compiled this list of conditions at the request of

the Health Resources and Services Administration (HRSA) of the federal government. The list, in turn, was endorsed by the March of Dimes. Expanded newborn screening is made possible by a relatively new screening technology known as Tandem Mass Spectrometry (abbreviated MS/MS). Using Tandem Mass Spectrometry, it is possible to test for many conditions at the same time using a single dried blood spot.

Tandem Mass Spectrometry is not an appropriate screening technology for all conditions currently being screened for in Colorado. Of the seven conditions on Colorado's existing bloodspot screening panel, only PKU (phenylketonuria) can be screened for using MS/MS. After July 1, this technology will be used to screen for PKU. The other six conditions on our panel will continue to be screened for using current methods.

The conditions being added to Colorado's panel are all "inborn errors of metabolism," meaning individuals with these conditions have an error in a metabolic pathway that inhibits their metabolization of certain substances found in food. These disorders fall into three categories: disorders of amino acid metabolism, organic acid metabolism, and fatty acid oxidation. These errors either cause unmetabolized substances to build up to toxic levels in the body, or in the case of fatty acid oxidation disorders, make it impossible for the body to break down fat to use for energy. Undiagnosed and untreated, these disorders lead to acute "metabolic crises" that in turn lead to coma and sometimes death. Those who survive these initial acute illnesses are often left with permanent disabilities and brain damage. As with the other conditions in our newborn screening program, children with these disorders appear healthy.

The conditions have, without exception, long and difficult-to-pronounce names. Because of this, most are referred to by acronyms. To add to the confusion, many of the conditions are known by more than one name – even as many as three or four different names – and there is no one widely accepted preferred nomenclature.

The most prevalent condition in the expanded panel is a disorder of fatty acid oxidation known as Medium Chain Acyl CoA Dehyrogenase Deficiency, commonly referred to as MCADD or MCAD and pronounced "em-cad." MCAD occurs with about the same prevalence as PKU (phenylketonuria), that is, one in every 15,000 births. In Colorado, we have found between one and five cases of PKU every year since mandated universal newborn screening went into effect in 1979. We will expect to find a similar number of cases of MCAD per year.

MCAD is easily and effectively treated. Because an individual with MCAD cannot break down fat from food or stored body fat for energy, he or she must never be allowed to "fast," meaning he or she must consume a sufficient amount of calories on a regular schedule. This can range from something as simple as always having a bedtime snack to immediate hospitalization for intravenous nutrition when he or she is too ill to take food by mouth and/or is vomiting. Other treatments include a high-carbohydrate, low-fat diet, and in some cases, dietary supplements.

The other conditions on the expanded panel are more rare than MCAD, and some are not as easily or effectively treated, but children with any of these conditions will benefit from early detection and intervention.

The added conditions on the expanded panel are individually extremely rare. However, as a group, it is estimated that they occur in the population at the rate of approximately 1:4,000 births.

Please refer to the following article, generously provided by the March of Dimes, for prevalence figures and short descriptions of the conditions on the expanded newborn screening panel.

For Further Information

For your convenience, we offer "one-stop shopping" at www.hcpcolorado.org!

Please visit the website for the Health Care Program for Children with Special Needs (HCP) of the Colorado Department of Public Health and Environment, at hcpcolorado.org. (Choose "Get to Know HCP," followed by "Screenings.") There you will find a wealth of information!

For each of the 29 conditions, we provide

- "ACT" sheets from the American College of Medical Genetics outlining ***immediate steps*** for the primary care provider to take upon receiving a positive screen for an infant in his or her practice
- Clinical intervention and specialty referral resources within Colorado
- Fact sheets providing general information about the disorder in terms easily understood by someone who is not medically trained
- A link to the site *Online Mendelian Inheritance in Man*, for more technical information, references, links to MEDLINE, and links to related resources at the National Center for Biotechnology Information
- Contact information for *local* support groups for families
- *More resources added every day!*

We would like to acknowledge and thank Colorado's State Genetics Coordinator, Steve Holloway, for creating this excellent web-based resource in advance of the implementation of expanded newborn screening.

The 29 Conditions on the Expanded Newborn Screening Panel

The following material has been reprinted (with very slight editing) from the March of Dimes' website, with the permission of the Colorado Chapter of the March of Dimes. (*marchofdimes.com/professionals/14332_15455.asp*)

The 29 disorders can be grouped into five categories:
Amino acid metabolism disorders
Organic acid metabolism disorders
Fatty acid oxidation disorders
Hemoglobinopathies
Others

Amino Acid Metabolism Disorders
This is a diverse group of disorders, with varying degrees of severity. Some affected individuals lack enzymes that are needed to break down the building blocks of protein called amino acids. Others have deficiencies in enzymes that help the body rid itself of the nitrogen incorporated in amino acid molecules. Toxic levels of amino acids or ammonia can build up in the body, causing a variety of signs and symptoms and even death.

PKU = Phenylketonuria
Incidence: greater than 1 in 25,000
Affected individuals have an inability to properly process the essential amino acid phenylalanine, which then accumulates and damages the brain. PKU can result in severe mental retardation unless detected soon after birth and treated with a special formula. Affected individuals must be kept on a low-phenylalanine diet at least throughout childhood and adolescence, and for females, during pregnancy.

MSUD = Maple syrup urine disease
Incidence: less than 1 in 100,000
This inborn error of metabolism can be lethal if unrecognized and untreated. There is a wide spectrum of clinical presentations, from mild to severe. Affected babies appear normal at birth but soon begin to have neurological symptoms. The disorder gets its name from the fact that the urine smells like maple syrup. Without dietary treatment, severely affected babies do not survive the first month; even those who do receive treatment may have irreversible mental retardation. Rapid diagnosis and treatment are major factors in survival and outcome. Treatment consists of a special low-protein diet, which will vary depending

on severity of symptoms, and sometimes, supplementation with a vitamin, thiamin. The diet must be continued indefinitely with frequent monitoring.

HCY = Homocystinuria
Incidence: less than 1 in 100,000
Individuals with this disorder lack an enzyme responsible for converting the amino acid homocysteine into cystathionine, which is needed for normal brain development. If undetected and untreated, homocystinuria leads to mental retardation, eye problems, skeletal abnormalities and stroke. Treatment consists of a special diet, one or more vitamins (B6 or B12) and other supplements (betaine).

CIT = Citrullinemia
Incidence: less than 1 in 100,000
Build-up in the body of citrulline and ultimately ammonia can begin during the newborn period or later in infancy. Without treatment, seizures, coma, brain damage and death can result. With early diagnosis and treatment, normal development is possible. Treatment includes a low-protein diet, medications to rid the body of amino groups to prevent ammonia build-up, and nutritional supplements.

ASA = Argininosuccinic acidemia
Incidence: less than 1 in 100,000
Most commonly, symptoms begin in the first few days of life, with build-up of argininosuccinic acid and ultimately ammonia resulting in brain swelling, coma and, sometimes, death. Survivors often suffer permanent neurological damage. Other affected children may develop symptoms later in infancy or childhood. Early diagnosis and treatment can be lifesaving; however, in spite of treatment, affected individuals remain susceptible to episodes of ammonia build-up, and most have some degree of brain damage. Treatment consists of a low-protein diet, avoiding fasting, medications to prevent ammonia build-up, nutritional supplements, and in some cases, liver transplant.

TYR I = Tyrosinemia type I
Incidence: less than 1 in 100,000
Due to absence of an enzyme, byproducts of the amino acid tyrosine, particularly a very toxic compound called succinylacetone, build up in the liver. Without treatment, symptoms generally begin in the first few weeks or months of life and progress to liver or kidney failure, nerve damage and death. Drug treatment, sometimes along with a low-protein diet, is very effective in preventing liver and kidney damage.

Organic Acid Metabolism Disorders
Each disease in this group of inherited disorders results from the loss of activity of an enzyme involved in the breakdown of amino acids, the building blocks of proteins, and other substances (lipids, sugars, steroids). When any of these chemicals is not properly broken down, toxic acids build up in the body. Without dietary treatment and prevention of acute episodes, these disorders can result in coma and death during the first month of life.

IVA = Isovaleric acidemia
Incidence: less than 1 in 100,000
This disorder is caused by an inability to process the amino acid leucine. The newborn form of the disorder often results in coma, permanent neurological damage and death. In other cases, symptoms develop later in infancy and childhood, frequently following an infectious illness. With early diagnosis and treatment, most children have normal development. Treatment includes a low-protein diet and nutritional supplements.

GA I = Glutaric acidemia type I
Incidence: greater than 1 in 75,000
Babies may develop normally for up to 18 months until something affects a child's health, such as a mild viral illness, which may trigger the onset of symptoms. Without prompt treatment, this can lead to brain damage, seizures, low muscle tone, cerebral-palsy-like symptoms and death within the first decade of life. Some affected babies also are born with an enlarged head (macrocephaly). Treatment can vary, but may include dietary protein restriction and supplementation with a nutrient called L-carnitine. With early diagnosis and prompt treatment of illness and fever, brain damage may be prevented.

HMG = Hydroxymethylglutaric aciduria or HMG-CoA lyase deficiency or 3-OH 3-CH3 glutaric aciduria
Incidence: less than 1 in 100,000
An inability to process the amino acid leucine leads to low blood sugar and accumulation of several organic acids, especially following illness or fasting. Without

treatment, the disorder can lead to brain damage, mental retardation, coma and death. Avoiding fasting and following a diet low in protein and fat and high in carbohydrates can lead to normal development.

MCD = Multiple carboxylase deficiency

Incidence: less than 1 in 100,000

This disorder is caused by a defect of an enzyme required to activate several biotin-dependent enzymes. Without these enzymes, lactic acid and other organic acids build up in the body. Without treatment, brain damage, coma and death can result. Symptoms usually begin between birth and 15 months of age, and may include skin rashes and hair loss. Early diagnosis and treatment with biotin allows normal growth and development.

MUT = Methylmalonic acidemia due to mutase deficiency

Incidence: greater than 1 in 75,000

A defect in the processing of four essential amino acids and other substances results in illness in the first week of life. Though severity of symptoms varies greatly, death during the first month of life and brain damage in survivors is common. Treatment includes a low-protein diet, vitamin B12 injections and nutritional supplements. Some children die during the first year of life or develop brain damage despite nutritional intervention.

Cbl A,B = Methylmalonic acidemia cblA and cblB forms

Incidence: less than 1 in 100,000

This inherited defect of vitamin metabolism can lead to build-up of acids in the blood and result in brain damage, seizures, paralysis, coma and death. Symptoms can begin as early as the first week of life, though a minority of affected individuals remains symptom-free. Treatment with vitamin B12 injections and a low-protein diet often prevents serious problems.

3MCC = 3-Methylcrotonyl-CoA carboxylase deficiency

Incidence: greater than 1 in 75,000

This defect in processing the amino acid leucine can lead to brain damage, seizures, liver failure and death in infancy or no symptoms at all into adulthood. Symptoms often develop following a childhood illness. Treatment with a low-protein diet and, in some cases, nutritional supplements may be helpful. (An abnormal result by newborn screening could be related to abnormal metabolites in the mother and not the baby. This will be clarified by further diagnostic testing of the infant.)

PROP = Propionic acidemia

Incidence: greater than 1 in 75,000

This defect in the processing of four essential amino acids leads to illness during the newborn period. Without treatment, brain damage, coma and death can result. Even with treatment, including a low-protein diet and nutritional supplements, some affected children suffer from developmental delays, seizures, abnormal muscle tone, frequent infections and heart problems.

BKT = Beta-Ketothiolase deficiency

Incidence: less than 1 in 100,000

Periodic episodes of acid build-up, often triggered by some childhood illness, can progress to coma, brain damage and death. These serious consequences are most often seen in infants. With early diagnosis and prompt intravenous treatment to keep blood sugar levels up and blood acid levels down during an illness, children can develop normally. Parents must be alert to early signs of illness. Additional treatments may vary, but can include avoidance of protein-rich diets and long-term treatment with bicarbonate.

Fatty Acid Oxidation Disorders

This group of disorders is characterized by inherited defects of enzymes needed to convert fat into energy. When the body runs out of glucose (sugar), it normally breaks down fat to support production of alternate fuels (ketones) in the liver. Because individuals with these disorders have a block in this pathway, their cells suffer an energy crisis when they run out of glucose. This is most likely to occur when an individual is ill or skips meals. Without treatment, the brain and many organs can be affected, sometimes progressing to coma and death.

MCAD = Medium-chain acyl-CoA dehydrogenase deficiency

Incidence: greater than 1 in 25,000

Seemingly well infants and children can suddenly develop seizures (caused by low blood sugar), liver failure, coma and death. Identifying affected children before they become ill is vital to preventing a crisis and averting these consequences. Treatment includes avoidance of fasting and nutritional supplements.

VLCAD = Very long-chain acyl-CoA dehydrogenase deficiency

Incidence: greater than 1 in 75,000
Symptoms can first appear at any age, from the newborn period through adulthood, but tend to be most severe in infants. Without treatment, affected infants often develop heart and liver failure and die during the first year of life. Treatment includes a high-carbohydrate/low-fat diet, nutritional supplements, avoidance of fasting and prolonged exercise.

LCHAD = Long-chain 3-OH acyl-CoA dehydrogenase deficiency

Incidence: greater than 1 in 75,000
Symptoms can begin soon after birth, resulting in heart, lung or liver failure and death. In other cases, symptoms such as low muscle tone; developmental delay; or heart, lung or liver failure may develop later in infancy or childhood, most likely following an illness. Early diagnosis and treatment effectively prevent life-threatening events, though some children may still develop symptoms. Treatment includes a high-carbohydrate/low-fat diet, nutritional supplements and avoidance of fasting. Women who are pregnant with fetuses with LCHAD are at increased risk of developing acute fatty liver of pregnancy and other pregnancy complications.

TFP = Trifunctional protein deficiency

Incidence: less than 1 in 100,000
A seemingly healthy infant can die suddenly of what appears to be sudden infant death syndrome. Other infants may develop low muscle tone, seizures, heart failure and coma, often following an illness. Treatment is based on strict avoidance of fasting, a low-fat diet and nutritional supplements.

CUD = Carnitine uptake defect

Incidence: less than 1 in 100,000
Due to a missing transporter, cells cannot bring in carnitine from the blood. Carnitine is needed for the transfer of fatty acids across the membranes of the mitochondria (cellular organelles that produce energy for the cell). Symptoms include episodes of hypoglycemia (low blood sugar) and sudden unexpected death in infancy. Older children may present with progressive heart failure. Early diagnosis and treatment with carnitine permits normal development.

Hemoglobinopathies

These inherited diseases of red blood cells result in varying degrees of anemia (shortage of red blood cells), serious infections, pain episodes and damage to vital organs. The symptoms are caused by abnormal kinds and/or amounts of hemoglobin, the main protein inside red blood cells that carries oxygen from the lungs and takes it to every part of the body. In the sickling disorders, an abnormal hemoglobin called HbS can cause some red blood cells to become stiff and abnormally shaped. The stiffer red blood cells can get stuck in tiny blood vessels, causing pain and sometimes organ damage. The severity of these disorders varies greatly from one person to the next.

Hb SS = Sickle cell anemia

Incidence: greater than 1 in 5,000; incidence among African-Americans 1 in 400
This blood disease can cause severe pain, damage to the vital organs, stroke and sometimes death in childhood. Young children with sickle cell anemia are especially prone to dangerous bacterial infections such as pneumonia and meningitis. Vigilant medical care and treatment with penicillin, beginning in infancy, can dramatically reduce the risk of these adverse effects and the deaths that can result from them. Affected babies should receive all regular childhood vaccinations (including hemophilus influenza B and pneumococcal vaccines) to help prevent serious bacterial infections. Additional treatments may vary according to severity of symptoms, but may include intermittent pain medications and regular blood transfusions.

Hb S/Th = Hb S/Beta-Thalassemia

Incidence: greater than 1 in 50,000
In this form of sickle cell anemia, the child inherits one sickle cell gene and one gene for beta thalassemia, another inherited anemia. Symptoms are often milder than for Hb SS, though severity varies among affected children. Routine treatment with penicillin may not be recommended for all affected children.

Hb S/C = Hb S/C disease

Incidence: greater than 1 in 25,000
Another form of sickle cell disease, in which the child inherits one sickle cell gene and one gene for another abnormal type of hemoglobin called HbC. As with Hb S/Th, this form is often milder the Hb SS and routine penicillin treatment may not be recommended.

Others
This mixed group of disorders includes some diseases that are inherited and others that are not genetic. This group of disorders varies greatly in severity, from mild to life threatening.

CH = Congenital hypothyroidism
Incidence: greater than 1 in 5,000
This thyroid hormone deficiency severely retards both growth and brain development. If detected soon after birth, the condition can be treated simply with oral doses of thyroid hormone to permit normal development.

BIOT = Biotinidase deficiency
Incidence: greater than 1 in 75,000
Biotinidase is the enzyme that recycles the vitamin biotin. An inherited deficiency of this enzyme may cause serious complications, including frequent infections, uncoordinated movement, hearing loss, seizures and mental retardation. Undiagnosed and untreated, the deficiency can lead to coma and death. If the condition is detected soon after birth, these problems can be completely prevented with daily oral doses of biotin.

CAH = Congenital adrenal hyperplasia
Incidence: greater than 1 in 25,000
CAH refers to a set of inherited disorders resulting from defects in the synthesis of hormones produced by the adrenal gland. In female infants, CAH sometimes results in masculinization of the genitals. Certain severe forms of CAH cause life-threatening salt loss from the body if undetected and untreated. Treatment includes salt replacement and hormone replacement.

GALT = Classical galactosemia
Incidence: greater than 1 in 50,000
Affected babies are missing the liver enzyme needed to convert galactose, a major sugar from the breakdown of lactose in milk, into glucose, another simple sugar that the body can use. Galactose then accumulates in and damages vital organs, leading to blindness, severe mental retardation, infection and death. Milk and other dairy products must be eliminated from the baby's diet for life. Though treatment dramatically improves the outlook for affected infants, there is still some risk of developmental delays.

HEAR = Hearing loss
Incidence: greater than 1 in 5,000, up to 3-4 per 1,000 newborns
Without early testing, most babies with hearing loss are not diagnosed until 2 or 3 years of age. By this time, they often have delayed speech and language development. Early diagnosis allows use of hearing aids by 6 months of age, helping prevent serious speech and language problems.

CF = Cystic fibrosis
Incidence: greater than 1 in 5,000
Cystic fibrosis is one of the most common inherited disorders in the United States. Abnormalities in the cystic fibrosis protein result in lung and digestive problems, and death at an average age of 30-35 years. Studies suggest that early diagnosis and treatment improves the growth of babies and children with CF. Treatment varies depending on severity of symptoms, but may include a high-calorie diet supplemented with vitamins and medications to improve digestion, respiratory therapy to help clear mucus from the lungs, and medications to improve breathing and prevent lung infections.

The NICU Patient – Special Considerations with Regard to Expanded Newborn Screening

Policy Change: Timing of First Screen for Neonatal Intensive Care Unit (NICU) Patients

NICUs should collect the initial newborn screen on babies by 72 hours of age.

Colorado's screening program has historically allowed Neonatal Intensive Care Units (NICUs) to wait up to one week (seven days) to screen premature and/or sick newborns. With the implementation of expanded newborn screening, metabolic specialists are adamant that this policy should be abandoned, and *all* newborns should have their initial newborn screening specimen collected at no later than 72 hours of age (the current recommendation for full term/well babies).

The conditions being added to Colorado's newborn screening panel are "inborn errors of metabolism." Some of these disorders can begin to make newborns ill as early as the first feeding. A baby may be in the NICU and begin to present symptoms of the disorder that are not specific enough to pinpoint the diagnosis, and certain medical interventions may actually worsen the baby's condition. If a NICU patient is not screened until seven days of age, results will not be available for several more days, and babies could die or suffer permanent neurological damage from the disorder before the results of the newborn screen are available. Shortening the interval until newborn screening results are available is vital for ensuring prompt diagnosis and treatment of children affected with these disorders. These rare conditions, if considered as a group, occur in the population at the rate of 1 in 4,000 births, so NICU staff will be encountering these disorders relatively frequently.

Screening Considerations: TPN

Once we begin expanded newborn screening, it is more important than ever to report on the lab slip if an infant is on total parenteral nutrition (TPN). Knowing that a baby was on TPN when a sample was collected is invaluable in helping the lab to interpret the screening results accurately and make the correct recommendation for further testing and clinical evaluation of the infant.

It is unavoidable that infants on TPN will have a higher rate of "false positive" screening results for certain of the conditions on the expanded panel, but with accurate information and experience, the screening program will be able to lessen this burden on providers.

As has been noted elsewhere in this article, to detect certain of the conditions on the expanded panel early enough to benefit the affected child, NICU patients ***must*** be screened within 72 hours of birth, regardless of whether or not they are on TPN.

Screening for Congenital Adrenal Hyperplasia (CAH) – Important Information and Reminders for Primary Care Providers

Colorado has been screening for Congenital Adrenal Hyperplasia (CAH) for more than five years. We have identified 21children with classic CAH and five children with one of the CAH variants. Before screening, 1 in 3 of these children would have experienced life-threatening illness before the disorder was recognized.

Classic salt-wasting CAH can lead to adrenal crisis in the first days of life accompanied by severe electrolyte disturbances, severe neurological damage or death. Classic virilizing CAH causes severe hormonal imbalances that can lead to gender missassignment, though salt-wasting may be subclinical. There are milder, variant forms of CAH that do not cause immediate problems in the neonatal period, but may interfere with normal growth and development, resulting in, for example, precocious puberty and severe short stature.

Colorado's newborn metabolic screening program has been highly successful in identifying infants with this disorder before the occurrence of serious consequences. However, as a result of lessons learned in the first years of screening, we have made some changes to our CAH screening procedures.

We have observed that a pattern of rising screening values over the two mandated newborn screens – no matter how slight the increase – is an important predictor of CAH. Therefore, the lab now considers any child who has a *higher value* on the *second screen* than on the first, to be a presumptive positive case of CAH, and recommends an *immediate* serum alpha-hydroxy-progesterone (17-OHP) test – the diagnostic test for CAH.

It follows that, because some children with CAH may have only a ***slightly abnormal*** or even a ***normal*** first newborn screen, it is vitally important to assure that ***all*** infants get their mandated second newborn screen and to be sure to rescreen any infant who has an abnormal screen for CAH, even if only mildly elevated.

Despite the screening laboratory's efforts to emphasize the ***urgency*** when calling out a presumptive positive screening result for CAH, there have been a number of affected children whose providers did not respond to the urgency of the situation. Children with salt-wasting classic CAH are in imminent danger of a life-threatening adrenal crisis within days of birth. Recommended testing and clinical follow-up should be done without delay. But remember, it may take several days to know the results of the diagnostic test for CAH. In any infant reported to have an abnormal CAH screen, even if only mildly elevated, it is important for the clinician to have a high index of suspicion for the clinical signs of the disorder. These include poor feeding, vomiting and weight loss. In the presence of these signs/symptoms, obtaining an electrolyte panel may be ***life-saving***.

Pediatric endocrinologists are under contract to the newborn screening program to provide consultation on appropriate and timely testing and clinical evaluation of children at risk for CAH. See the resource section in this newsletter for their contact information.

Congenital Hypothyroidism Found on the Second Newborn Screen

Colorado's experience with finding cases of congenital hypothyroidism on the second newborn screen (after a normal first screen) has been published in the *Journal of Pediatric Endocrinology and Metabolism* (19:1, 31-38, January, 2006) by Dr. Aristides Maniatis, et al. The conclusion drawn in the article is that without a mandated second newborn screen, one case of congenital hypothyroidism would be missed in every 11,111 babies screened in Colorado. With Colorado's birth rate at 70,000, this means that, without a mandated second newborn screen, we would be missing half a dozen infants with this disorder every year. This number is higher than had been previously reported by other newborn screening programs. This research was also presented at the National Endocrine Society meeting in 2005 and received the Abbott Thyroid Research Award.

Filling Out the Newborn Screening Lab Slip

It is ***vitally*** important to fill out lab slips as ***accurately***, ***completely*** and ***legibly*** as possible. The lab needs accurate information on factors such as birth weight, blood transfusions and TPN to guarantee the reliability of the results. The ability to reach the appropriate provider ***quickly*** and ***unerringly*** could be ***a matter of life and death*** to a baby.

Introducing Emily Fields, Colorado's Newborn Hearing Screening Follow-up Coordinator

In January 2006, Colorado's Newborn Hearing Screening Program was very lucky to hire Emily Fields, MS, as its new follow-up coordinator.

Emily is a Colorado native and a graduate of the University of Colorado.

Emily is a board-certified genetic counselor. She came to the Hearing Screening Program from the Hereditary Cancer Clinic at the University of Colorado Health Sciences Center.

Fifty percent of hearing loss is genetic. Emily's education and expertise in genetics will allow the hearing screening program to expand in new directions.

And, in honor of her new position, Emily has adopted a dog that is deaf – the lovely Lily.

Emily can be reached at 303-692-2349 or by e-mail at emily.fields@state.co.us.

In Appreciation: Joanne McConville

In 2001, Joanne McConville, MBA, was hired as Colorado's first full-time newborn hearing screening follow-up coordinator.

In her four years in that position, Joanne helped to create a system to follow up on Colorado babies with abnormal newborn hearing screens and assure diagnosis and intervention for affected children by six months of age. It has been proven that children who are deaf or hard of hearing who are diagnosed and receive intervention by six months of age develop speech and language on a par with children with normal hearing. To achieve this goal, Joanne worked intensively with hospital nursery staff, families, primary care providers, state health department staff, audiologists and early childhood specialists to create a system and a network of professionals dedicated to providing optimal outcomes for children who are deaf or hard of hearing.

Joanne observed that screening rates among home-birthed infants and Hispanic infants born in certain hospitals lagged behind other groups. Joanne worked with lay midwives to increase the number of home-birthed infants who receive a newborn hearing screening – increasing that number from essentially none to almost 25% of all babies born at home. She initiated extensive outreach with clinics providing primary care to the Hispanic community and attended Hispanic health fairs to provide on-site newborn hearing screenings; these efforts have significantly increased screening and rescreening rates in Colorado's burgeoning Hispanic population.

Joanne has moved on, taking another position with the Colorado Department of Public Health and Environment. We would like to take this opportunity to thank Joanne and to acknowledge her for her professionalism, her enthusiasm, her initiative, her creativity, her dedication, and the many ways in which she contributed to Colorado remaining an international leader in newborn hearing screening.

Newborn Hearing Screening in Colorado: 2006 Update

Albert Mehl, MD
Chairman,
Colorado Infant Hearing Advisory Committee

Since the early 1990s, Colorado has been a leader in recognizing the importance of and in providing universal newborn hearing screening. Colorado physicians were at the forefront in demonstrating the feasibility of screening every newborn for hearing loss, using new technologies that allowed an assessment of hearing long before infants could respond to a behavioral hearing test. Soon thereafter, Colorado researchers published the dramatic improvements in language outcomes when hard-of-hearing children were identified in infancy, rather than the typical delay of two years or more in the absence of newborn screening. Currently more than 95 percent of Colorado newborns have hearing screening performed prior to hospital discharge.

Despite our successes in Colorado, there is much work yet to be done. Not all primary care providers realize that potential hearing loss in newborns, just like an abnormal screen for phenylketonuria or congenital hypothyroidism, is a "developmental emergency." In looking toward the goal of improving outcomes, primary care providers should remember the numbers "One, Three and Six."

Before One Month: Every newborn should complete hearing screening and rescreening as soon as possible, but always by ***one*** month of age. Even babies born at home or in a birthing center rather than a hospital should be offered early hearing screening. And infants who fail just one rescreening as an outpatient should be referred directly to a qualified audiologist for a comprehensive evaluation.

Before Three Months: Every newborn who fails initial screening, and then fails a single outpatient rescreening, should complete a comprehensive audiology evaluation as soon as possible, but always before ***three*** months of age.

Before Six Months: Every infant identified with congenital hearing loss should begin a coordinated early intervention program as soon as possible, but always by ***six*** months of age. Because 90 percent of infants with congenital hearing loss have some residual hearing, amplification with a hearing aid is a mainstay of this early treatment. Even infants who are profoundly deaf will dramatically benefit from the early use of sign language, as well as being potential candidates for a cochlear implant.

The primary care provider (PCP) is a critical component to the success of a statewide program for newborn hearing screening and intervention. The PCP can assure that families understand the importance of returning promptly for reassessment. The primary care provider coordinates referrals to a qualified audiologist and to an otolaryngology specialist, and must be familiar with the experience level of these professionals and the special technologies required of them when assessing newborns. For infants with confirmed hearing loss, the primary care provider must arrange for prompt and aggressive intervention to assure a good outcome for the child and must reinforce with the family the importance of using the hearing aid during every waking hour of the day. The primary care provider must assure that the infant with confirmed hearing loss is examined by a pediatric ophthalmologist and assessed for any co-morbidities of vision. And finally, as half or more of all newborn hearing loss has a genetic basis, families should be offered an evaluation by a qualified genetic counselor.

Problems Persist

Many primary care providers do <u>not</u> realize:

- Although more than 95 percent of infants born in hospitals in Colorado have screening performed prior to discharge, many who are born at home or in birthing centers may never have hearing screening performed.
- Twenty percent of infants who fail their initial hearing screening never return for the recommended rescreening to diagnose or exclude hearing loss.
- Newborn hearing loss is a "developmental emergency," and primary care providers play an important role in assuring recall of infants for diagnostic testing and rapid enrollment in early intervention programs.
- An infant can be fit with a hearing aid as early as one month of age.
- An infant's hearing can be evaluated even when middle ear effusions are present, using bone conduction techniques.
- Ninety percent of newborns identified with hearing loss have only partial hearing loss; these infants will benefit dramatically from receiving early amplification.
- Assessing for hearing loss and fitting tiny babies with hearing aids requires special skill and equipment and should only be performed by audiologists with appropriate training.

- Even when profound hearing loss is identified, language can be acquired at a normal pace by teaching sign language to the family and the infant, and the infant can be expected to be signing words by the first birthday, just as with spoken language.

- Every infant identified with hearing loss should be referred to a pediatric ophthalmologist to evaluate the child for co-morbidities of vision.

- A genetics evaluation should be offered to the family of every baby diagnosed with hearing loss, as more than half of all newborn hearing loss has a genetic basis.

To summarize, what can you, as the primary care provider, do to assure success for our Colorado Infant Hearing Screening Program and optimal outcomes for your patients?

▶ Work with your local audiologist to learn more, and watch for continuing educational opportunities related to newborn hearing screening.

▶ Make sure that infants who do not pass their hearing screening in the hospital are reassessed as early as possible and always before one month of age.

▶ If an infant does not pass a single outpatient rescreening, make sure the child completes a comprehensive audiology evaluation as soon as possible and always before three months of age, with an audiologist with pediatric experience.

▶ For infants with confirmed hearing loss, assist the family in beginning early intervention as soon as possible and always before six months of age.

For more information, contact:

Albert Mehl, MD
Chairman
Colorado Infant Hearing Advisory Committee
720-536-7575
albert.mehl@kp.org

Vickie Thomson
Colorado Early Hearing Detection and Intervention Coordinator
303-692-2458
vickie.thomson@state.co.us

Evaluating an Early Hearing Detection and Intervention (EDHI) Program Using "Sound" Data

A newborn hearing screening program is only successful if those infants who fail the screen receive appropriate and timely follow-up. The Colorado Department of Public Health and Environment's Health Care Program for Children with Special Needs (HCP) is responsible, by legislation, to assure that a comprehensive system, from screening through diagnosis and early intervention, exists. As our database continues to improve, we are now able to analyze our data to identify the successes and gaps in our system. The EHDI staff has started looking at the data beyond the screening stage.

In our initial analysis, we discovered that some populations of infants are more likely to be missed for hearing screening. The data show that infants born in the NICU missed their initial screen more often than any other population of newborns. Further analysis will help us determine which infants are missed and how to implement a safety net to assure they receive a screen. All NICU infants are at greater risk for hearing loss and should receive this important screening test. We also found that Latino infants were generally at higher risk to not receive an outpatient follow-up screen. However, this is not always true and seems to depend greatly on birth hospital. For example, Denver Health Medical

Center has a "medical home" system and they do a remarkably thorough job of getting their Latino infants to return for rescreening.

In an article published in *Pediatrics*,[1] the authors suggest that a two-stage screening with otoacoustic emissions (OAE) followed by automated auditory brainstem response (AABR) may be missing 23% of mild congenital hearing loss cases, and specifically that screening using AABR misses these infants. Colorado has been screening newborns since 1992 with AABR and we have not found this to be the case. Additionally, the authors did not use a control group. Therefore, it could not be firmly established that those infants who passed their newborn hearing screen and were later identified with hearing loss were truly missed by screening or were among a group of infants that present with late onset hearing losses from causes such as asymptomatic perinatal cytomegalovirus (CMV) infection.[2] The incidence of late onset hearing loss due to CMV reaches 15.4% by 72 months after birth in cases of congenital infection. The incidence of perinatal CMV is substantial, 0.5-1.5% of live births. Ninety per cent of these perinatal infections are asymptomatic in infants, making CMV a significant consideration when considering late onset infant hearing loss that is likely to be missed by newborn infant hearing screening.[3]

Our data demonstrate that hospitals using OAE *only* have very high false positive rates (greater than 10%) when compared to those that use the two-stage protocol of OAE followed by AABR (less than 4%). In addition, those hospitals that use OAE alone (as Johnson et al. prefer) are not identifying a greater number of congenital hearing losses than those that use the two-stage OAE/AABR protocol or use AABR exclusively.

The Colorado Infant Hearing Advisory Committee recommends that hospitals select the screening protocol that works best for their population. Audiology Regional Coordinators (their contact information is provided in the resource section of this newsletter) are available statewide to help hospitals identify the technology and protocol that works best for the hospital and the community. Keep in mind that both technologies will miss some degrees and some types of hearing loss and there will be infants who pass their newborn hearing screen who will have a late onset of hearing loss. Whenever a parent has a concern or there is a high risk factor (e.g., family history, craniofacial anomalies, in-utero infections, stigmata associated with a syndrome, head trauma or recurrent ear infections for at least three months) the child should be referred to an audiologist with pediatric expertise. *Hearing can be tested at any age!*

The Joint Committee on Infant Hearing (with representatives from the American Academy of Pediatrics, the American Academy of Otolaryngology and others) will soon publish a position statement to provide additional guidance on recommended protocols and follow-up for newborn hearing screening programs. Automated auditory brainstem response (AABR) will be recommended as the preferred screening method for all NICU infants, in order to rule out the disorder called auditory neuropathy. The Colorado Infant Hearing Advisory Committee Guidelines also make this recommendation. The Guidelines are available at www.hcpcolorado.org.

Over the next year, we will be doing an in-depth analysis of the following:

- What factors contributed when infants who failed the newborn hearing screen did not receive a diagnosis by three months of age?
- What factors were present when infants passed their newborn screens but were later identified with hearing loss?

HCP has been working with the federal Centers for Disease Control and Prevention (CDC) to develop a Family Satisfaction Survey to evaluate the EHDI program in Colorado. Fifteen hundred surveys were sent to families whose infants passed their newborn hearing screen, fifteen hundred surveys were sent to families whose infant failed the screen at discharge and subsequently passed an outpatient screen, and 250 surveys were sent to those families whose infants were confirmed with a permanent hearing loss between 2002 and 2004. The results of the surveys are currently being analyzed. HCP will use these data to evaluate the current EHDI system, with the goal of improving the services we provide for the children and families that we serve.

[1]Johnson, J.L., White, K.R., Widen, J.E., Gravel, J.S., James, M., Kennalley, T., Maxon, A.B., Spivak, L., Sulivan-Mahoney, M., Wohr, B.R., Weirather, Y., Hostrum, J. A multicenter evaluation of how many infants with permanent hearing loss pass a two-stage otoacoustic emissions/automated auditory brainstem response newborn hearing screening protocol. *Pediatrics* 2005; 116:663-672.

[2]Fowler K.B., Dahle A.J., Boppana S.B., Pass R.F. Newborn hearing screening: Will children with hearing loss caused by congenital cytomegalovirus infection be missed? *J Pediatr* 1999; 136:60-4.

[3]Demmler G. Summary of a workshop on surveillance for congenital cytomegalovirus disease. *Rev Infect Dis* 1991; 13:315-29.

How to Obtain Newborn Screening Results

Hearing Screening Results
Contact Emily Fields, Follow-up Coordinator
Newborn Hearing Screening Program
by phone, fax or e-mail
303-692-2349
FAX 303-753-9249
emily.fields@state.co.us

Metabolic Screening Results
The Laboratory charges fees for all requests for information falling under the State of Colorado Open Records Act. This includes requests for copies of test results that have been previously reported to the submitting facility and copies of test results requested to be sent to other than the submitting facility.

The lab charges 50¢ per page for copies and $1.00 per page for faxed documents. The lab requires receipt of a parental/patient release authorization prior to releasing test information to other that the original submitting agency for test results already reported to the referring hospital, laboratory or physician. The signed release must accompany the request.

In order to serve you more efficiently, the lab prefers that you use its request form to request newborn screen results. This form can be found online at www.cdphe.state.co.us/lr/NBS/NBSResultsRequestForm.doc or you may request from the lab by phone at 303-692-3670. Return completed forms by mail to the attention of Eric Struxness, Laboratory Services Division, 8100 Lowry Blvd., Denver, CO, 80230, or by fax at 303-691-4057. Results will be sent to you by mail or fax within 72 hours of receipt. If you need any further assistance, please call 303-692-3056.

How to Order Brochures and Lab Slips

To order newborn *metabolic* screening lab slips and parent education brochures (in English and Spanish), call 303-692-3670.

To order newborn *hearing* screening brochures (in English and Spanish), call 303-692-2370.

HCP: Connecting Kids with Care

The Newborn Metabolic Screening Follow-up Program and the Newborn Hearing Screening Program are programs of the Health Care Program (HCP) for Children with Special Needs at the Colorado Department of Public Health and Environment.

What can HCP do for your pediatric patient with special health care needs?

There are more than 225,000 children in Colorado with special health care needs. Special health care needs can be physical, emotional or behavioral. Conditions can be chronic or acute. These conditions can be easy to see or not seen at all. In addition to the conditions found through newborn metabolic screening and newborn hearing screening, HCP helps families of children with, or at risk for, such conditions as asthma, autism, cancer, cerebral palsy, cleft palate, developmental disabilities, diabetes, traumatic brain injury and vision problems, to name just a few.

HCP offices are located throughout Colorado, serving these children from birth to age 21. HCP helps these children, their families and their health care providers. HCP offers help in a crisis or help for the long term. The Health Care Program for Children with Special Needs

- provides community-based screenings, evaluations and clinics
- provides service referrals and care coordination for complex medical situations
- connects families with other families in the local community for support
- coordinates primary and specialty medical care for patients
- is available to providers to discuss solutions to difficult care situations and provide information and support as needed
- helps families find insurance and other financial assistance

There is an HCP office in your community. To locate your local office, refer to the list provided with this newsletter, visit the HCP website at www.hcpcolorado.org, or call 303-692-2370.

Universal Vision Screening: Update

Paula Hudson, PhD
Health Care Program for Children with Special Needs, Colorado Department of Public Health and Environment

Since 2004, The Health Care Program for Children with Special Needs of the Colorado Department of Public Health and Environment has been collecting data to ascertain the number of Colorado children between 12 months and 60 months of age who are receiving vision screening. The data collected have shown that approximately one half of all Colorado children (140,000 children) are screened for vision disorders before they enter school. (Approximately 70,000 children are born in Colorado every year.) The majority of these children are three to four years old. Rarely are younger children screened. Nationally, early vision screening (defined as a basic battery of screening tests, visual examination and history) is recognized as a vital service to young children. Many states have begun early screening programs for children as young as 18 months.

InfantSEE is a national program that became available in Colorado in 2005. It provides for all children younger than 12 months to have a basic vision evaluation by an optometrist or ophthalmologist free of charge. The participating optometrists and ophthalmologists can be found

on the web site (InfantSEE.com). Just enter the appropriate ZIP code.

Screening services for children older than 12 months are not well organized. Pediatricians and family physicians carry the majority of the burden, with schools, Child Find, Headstart and private entities also doing some vision screening.

The state health department is making a concerted effort to systematize the screening efforts in Colorado by establishing best practices and suggesting uniform screening procedures.

Vision disorders and blindness have been recognized as reportable disorders by the federal government. Thus, the Colorado Department of Public Health and Environment is collecting and will continue to collect pertinent data. These data will reveal the state of vision health in the population of children 12 months to 60 months of age in Colorado.

Do You Know About the Traumatic Brain Injury Trust Fund?

More than 3,000 Coloradoans sustain a traumatic brain injury (TBI) every year. A TBI – also called a head injury or a closed head injury – is defined as damage to the brain caused by outside force, such as a fall or a motor vehicle accident. It does not include brain injury caused by congenital anomalies, degenerative diseases, surgical interventions or anoxia.

The Colorado Traumatic Brain Injury Trust Fund was created in 2002. The TBI Trust Fund does not charge for its services; it is funded by surcharges on fines for drunk driving and speeding. The TBI Trust Fund provides care coordination, and more, for a period of one year. Care coordinators help each client develop a care coordination plan and find the best care and support. The coordinators facilitate cooperation among the patient, family, primary care provider and specialists. The Trust Fund can pay for some client services if the client does not have other health or rehabilitation benefits. Examples of such services include speech therapy, cognitive therapy, physical rehabilitation and respite care, among others. The Trust Fund *cannot* pay for inpatient hospital care, institutional care and medications. The Trust Fund provides services for children and adults. Children ages birth through 21 receive TBI Trust Fund services through The Health Care Program for Children with Special Needs (HCP) of the Colorado Department of Public Health and Environment. (Adults over 21 receive services from a different agency.)

To use the TBI Trust Fund, a client must be a legal Colorado resident and have proof of a TBI that produces a partial or total disability. The Trust Fund *may* pay for a diagnostic assessment in some cases. The client must have no other health or rehabilitation benefits that will pay for the services being provided. For additional information, visit the TBI Trust Fund website at www.tbicolorado.org.

All applications for Trust Fund services are processed by the Brain Injury Association of Colorado (BIAC). Contact BIAC to obtain an application or for further information. Call toll free at 1-888-331-3311 or visit the website at www.biacolorado.org.

If a child does not qualify for the TBI Trust Fund, families can still contact HCP for basic resource and referral information, at 303-692-2370, or toll free at 1-800-886-7689, ext. 2370.

If you have questions or comments about anything in this newsletter, please contact the editor Laura Taylor, at 303-692-2425 or by e-mail at laura.taylor@state.co.us.

Resources in Newborn Screening and Genetics for *Professionals*

The Health Care Program for Children with Special Needs (HCP) website at www.hcpcolorado.org offers "ACT" sheets from the American College of Medical Genetics outlining ***immediate steps*** for the primary care provider to take upon receiving a positive newborn screen for an infant, clinical intervention and specialty referral resources within Colorado, fact sheets providing general information about the disorder in terms easily understood by someone who is not medically trained, and other resources.

The Newborn Screening Laboratory website at www.cdphe.state.co.us/lr/NBS/ns_hom.asp provides rules and regulations for newborn screening, frequently asked questions about newborn screening, illustrated instructions for specimen collection and handling, instructions for ordering screening supplies, and instructions on how to obtain copies of newborn screening results.

The Mountain States Genetics Network (MoSt GeNe) maintains a website at www.mostgene.org. There you can find the Network's publications, online and free of charge; links to other reputable providers of genetics information; and other resources.

National Newborn Screening and Genetics Resource Center, 1912 W. Anderson Lane, #210, Austin, TX 78757, 512-454-6419, http://genes-r-us.uthscsa.edu

The American Academy of Pediatrics (AAP), 141 Northwest Point Blvd., Elk Grove Village, IL 60007-1098, 847-434-4000, FAX 847-434-8000, www.aap.org

The March of Dimes, www.marchofdimes.com

Clinical and Laboratory Standards Institute (CLSI) [formerly The National Committee for Clinical Laboratory Standards (NCCLS)] sells a video on newborn screening specimen collection entitled *Making a Difference through Newborn Screening*: *Blood Collection on Filter Paper*, 1999, $175 non-member price. This price includes both the video and *Blood Collection on Filter Paper…* (described next), order code LA04-A3-V. *Blood Collection on Filter Paper for Neonatal Screening Programs, Approved Standard – 4th Ed* (2003), $120 non-member price, order code LA4-A4. Contact info: 940 West Valley Road, Suite 1400, Wayne, PA 19087-1898; order toll free at 1-877-447-1888 or fax to 610-688-6400; customerservice@clsi.org

Visual Aids to Assist in Specimen Collection. Two full-color wall charts on specimen collection: *Newborn Screening Blood Specimen Collection and Handling Procedure* and *Simple Spot Check* (invalid specimens and their causes) are available at no charge from Whatman (formerly Schleicher & Schuell), 1-800-WHATMAN or 973-245-8300, fax: 973-245-8324, info@whatman.com. These two charts also are available as PDF files on the newborn screening laboratory's website at www.cdphe.state.co.us/lr/NBS/ns_hom.asp.

Guidelines for Infant Hearing Screening, Audiological Assessment, and Intervention (2004). These guidelines for primary care physicians, audiologists and others involved in infant hearing screening are published by Colorado's Infant Hearing Advisory Committee and are available as a PDF file at hcpcolorado.org. To obtain a printed copy, call 303-692-2370.

Resources for *Families*

Health Care Program for Children with Special Needs (HCP), www.hcpcolorado.org, 303-692-2370

Save Babies Through Screening, www.savebabies.org, 1-888-454-3383

newbornscreening.info

Parent to Parent of Colorado - Connecting families of children with disabilities or special health care needs in communities across Colorado, www.p2p-co.org, 1-877-472-7201; assistance in Spanish available

Family Voices - Families and friends who care for and about our children with special health care needs, www.familyvoicesco.org, 1-800-881-8272; for assistance in Spanish, please contact El Grupo VIDA (see below)

El Grupo VIDA - Parents of children with a disability or a special need benefit from the emotional support they receive from other parents who have the same experience, 303-904-6073, elgrupovida.org; assistance in Spanish available.

Family Healthline - A statewide telephone helpline and referral service that helps Coloradans access Colorado health programs, including indigent medical care, vision, dental, mental health and nutrition programs; emergency shelter; domestic violence; victim assistance; legal aid; housing assistance; and others. Call toll free 1-800-688-7777. Assistance is available in English and Spanish and for the hearing impaired.

My Baby's Hearing - www.babyhearing.org, information on hearing screening in English and Spanish

Colorado Families for Hands and Voices, support services for families of children with hearing loss, 303-300-9763, toll-free 1-866-422-0422, www.handsandvoices.org

Consultants for Assistance with Screening, Diagnosis and Medical Management

Newborn Screening Laboratory

for lab results and assistance with screening

Daniel Wright, Supervisor
Newborn Screening Laboratory
Colorado Department of Public Health and Environment
Laboratory Services Division
Mailing address for specimens and correspondence: P.O. Box 17123, Denver, CO 80217
Laboratory location: 8100 Lowry Blvd., Denver
303-692-3670
Fax: 303-691-4008

Newborn *Metabolic* Screening Follow-up

for assistance with follow-up of abnormal screens

Laura Taylor, Follow-up Coordinator
Newborn Metabolic Screening Program
Health Care Program for Children with Special Needs
Colorado Department of Public Health and Environment
4300 Cherry Creek Drive South
Denver, CO 80246-1530
303-692-2425
Fax: 303-753-9249
E-mail: laura.taylor@state.co.us

Newborn *Hearing* Screening Follow-up

Emily Fields, MS
Follow-up Coordinator
Newborn Hearing Screening Program
Health Care Program for Children With Special Needs
Colorado Department of Public Health and Environment
4300 Cherry Creek Drive South - A400
Denver, Colorado 80246-1530
303-692-2349
Fax: 303-753-9249
E-mail: emily.fields@state.co.us

Sickle Cell Disease • Abnormal Hemoglobin

Dr. Kathryn Hassell
Dr. Rachelle Nuss
Donna Holstein, RN
Sickle Cell Treatment and Research Center
University of Colorado Health Sciences Center, Box C-222
4200 E. Ninth Avenue
Denver, CO 80262
303-372-9070
Fax: 303-372-9161

Congenital Hypothyroidism • Congenital Adrenal Hyperplasia (CAH)

Dr. Michael Kappy
Dr. Philip Zeitler
Dr. Sharon Travers
Dr. Jennifer Barker
Dr. Kristen Nadeau
Dr. Francis Hoe
Division of Pediatric Endocrinology
The Children's Hospital, Box B-265
1056 E. 19th Avenue
Denver, CO 80218
303-861-6128
Fax: 303-864-5679

Dr. Clifford Bloch
Dr. Sunil Nayak
Dr. Aristides Maniatis
Pediatric Endocrine Associates
8200 East Belleview Avenue, # 510-East
Greenwood Village, CO 80111
303-783-3883
Fax: 303-783-3800
www.denverpedendo.com

Cystic Fibrosis

Clinical Consultation
Dr. Frank Accurso
Cystic Fibrosis Center
The Children's Hospital, Box B-395
1056 E. 19th Avenue
Denver, CO 80218
303-837-2522
Fax: 303-837-2924

Sweat Chloride Testing
*A diagnostic sweat chloride test may be obtained **at no charge** at University Hospital in Denver for infants who have had **two** abnormal newborn screens for Cystic Fibrosis*. Contact: University Hospital Laboratory Client Services, 303-372-0522

PKU • Galactosemia • Biotinidase Deficiency • Amino Acid Metabolism Disorders Organic Acid Metabolism Disorders • Fatty Acid Oxidation Disorders

Dr. Janet Thomas
Dr. Johan Van Hove
Dr. Renata Gallagher
The Inherited Metabolic Diseases Clinic
The Children's Hospital, Box B-153
1056 E. 19th Avenue
Denver, CO 80218
303-861-6847
Fax: 303-764-8024

Dr. Stephen Goodman
Chief, Clinical Genetics and Metabolism
Biochemical Genetics Laboratory
MS-8313 – P.O. Box 6511
Aurora, CO 80045-0511
303-724-3825
303-724-3826 (main lab #)
FAX 303-724-3827

Health Care Program for Children with Special Needs (HCP)
Audiology Regional Coordinators

Regional Office	*Discipline Contact and Address*	*Contact Information*	*County(ies)*
Boulder	Sally Specht Boulder County Public Health 529 Coffman, Suite 200 Longmont, CO 80501	Phone: 303-678-6172 Fax: 303-441-1452 Email: sspecht@co.boulder.co.us	Boulder
Denver	Trudy Frederics 9189 Seven Arrows Trail Lone Tree, CO 80124	Phone: 303-788-8351 Fax: 303-649-1875 Email: trudyfrederics@yahoo.com	Denver
El Paso	Kathy Sera Budney 175 S Union Blvd Suite 330 Colorado Springs, CO 80910	Phone: 719-442-6984 Alt. Phone: 719-495-3617 Fax: 719-442-6985 Email: kbudney@divide.net	El Paso, Teller
Jefferson	Trudy Frederics 9189 Seven Arrows Trail Lone Tree, CO 80124	Phone: 303-788-8351 Alt. Phone: 303-649-1873 Fax: 303-649-1875 Email: trudyfrederics@yahoo.com	Broomfield, Clear Creek Park, Gilpin Jefferson
Larimer	POSITION VACANT	Phone: 000-000-0000 Fax: 000-000-0000 Email:	Larimer
Northeast	Linda Jelden 315 South Reynolds Drive Holyoke, CO 80734	Phone: 970-854-2593 Fax: 970-522-1412 Email: ljelden@pctelcom.coop	Logan, Morgan Phillips, Sedgwick Washington, Yuma
Northwest	John Burke P. O. Box 770042 Steamboat Springs, CO 80477	Phone: 970-879-7286 Fax: 970-870-1326 Email: jburke1051@hotmail.com	Grand, Moffat, Routt Rio Blanco
Pueblo	Andrea Lucas Greenwood Ear Nose and Throat Specialists 1619 N Greenwood Ste 309 Pueblo, CO 81003	Phone: 719-544-7115 Fax: 719-442-6985 Email: andraelucas@yahoo.com	Pueblo

Regional Office	*Discipline Contact and Address*	*Contact Information*	*County(ies)*
South Central	Daria Stakiw - **Lake County Only** 433 Stagecoach Lane New Castle, CO 81647	Phone: 970-984-3270 Fax: 970-248-6972 Email: dariastakiw@yahoo.com	Alamosa, Chaffee Conejos, Costilla Custer, Fremont Huerfano, Lake Las Animas, Mineral Rio Grande Saguache
	Lara Ciasulli San Luis Valley BOCS PO BOX 1198 Alamosa, CO 81101	Phone: 719-589-5851 Fax: 719-589-5007 Email: lciasulli@slvbocs.org	
Southeast	Ty Squires 28095 E Everett Pueblo, CO 81006	Phone: 719-542-1760 Fax: 719-383-3060 Email: t_squiresms@hotmail.com	Baca, Bent Cheyenne, Crowley Kiowa, Kit Carson Lincoln, Otero Prowers
Southwest	Eileen Goebel 799 E 3rd St Durango, CO 81303	Phone: 970-375-2369 Fax: 970-375-9054 Email: avahear@frontier.net	Archuleta, Dolores La Plata Montezuma San Juan
Tri-County Health Department	Jill Boice 876 Downing Denver, CO 80218	Phone: 303-863-7580 Fax: 303-220-9208 Email: jill@booksandcats.net	Adams, Arapahoe Douglas, Elbert
Weld	Deanna Meinke University of Northern Colorado Gunter Hall Office 1500 Campus Box 140 Greeley, CO 80634	Phone: 970-351-1600 Fax: 970-351-2974 Email: deanna.meinke@unco.edu	Weld
Western Slope	Daria Stakiw 433 Stagecoach Lane New Castle, CO 81647	Phone: 970-984-3270 Fax: 970-248-6972 Email: dariastakiw@yahoo.com	Delta, Eagle Garfield, Gunnison Hinsdale, Mesa Montrose, Ouray Pitkin, San Miguel Summit
	Richard Hartman 2569 Silver Oak Court Grand Junction, CO 81501	Phone: 970-245-1740 Fax: 970-245-1740 Email: dhartma@bresnan.net	

State HCP Office

Vickie Thomson, MA, Audiology Consultant
Colorado Department of Public Health and Environment
Health Care Program for Children with Special Needs, PSD-HCP-A4
4300 Cherry Creek Drive South
Denver, CO 80246-1530

Phone: 303-692-2458
Fax: 303-782-5576
Email: vickie.thomson@state.co.us

Health Care Program for Children with Special Needs (HCP)
HCP Regional Office Team Leaders

Regional Office	*Discipline Contact and Address*	*Contact Information*	*County(ies)*
Boulder	Mary Adler Boulder County Public Health 529 Coffman, Suite 200 Longmont, CO 80501	Phone: 303-678-6137 Fax: 303-678-6125 Email: madler@co.boulder.co.us	Boulder
Denver	Marti Potter Health Care Program for Children with Special Needs; Denver Regional Office 1001 Yosemite Street MC#1701 Denver, CO 80230	Phone: 720-956-2057 Fax: 720-956-2159 Email: marti.potter@dhha.org	Denver
	Molly Benkert Health Care Program for Children with Special Needs; Denver Regional Office 1001 Yosemite Street MC#1701 Denver, CO 80230	Phone: 720-956-2150 Fax: 720-956-2159 Email: mbenkert@dhha.org	
El Paso	Ann Stager El Paso County Department of Health and Environment 301 South Union Boulevard Colorado Springs, CO 80910-3123	Phone: 719-578-3283 Fax: 719-578-3234 Email: annstager@epchealth.org	El Paso, Teller
	Jonni Hulse El Paso County Department of Health and Environment 301 South Union Boulevard Colorado Springs, CO 80910-3123	Phone: 719-578-3226 Fax: 719-578-3234 Email: jonnihulse@epchealth.org	
Jefferson	Carolyn Kwerneland Jefferson County Department of Health and Environment 260 South Kipling Street Lakewood, CO 80226	Phone: 303-239-7014 Fax: 303-239-7088 Email: ckwernel@jeffco.us	Broomfield, Clear Creek Gilpin, Park, Jefferson
Larimer	Ginger Hillyard Larimer County Department of Health and Environment 1525 Blue Spruce Drive Fort Collins, CO 80524-2004	Phone: 970-498-6732 Fax: 970-498-6772 Email: ghillyard@larimer.org	Larimer
	Averil Strand Larimer County Department of Health and Environment 1525 Blue Spruce Drive Fort Collins, CO 80524-2004	Phone: 970-498-6760 Fax: 970-498-6772 Email: strandaa@co.larimer.co.us	
Northeast	Paul Gillenwater Northeast Colorado Health Department 228 West Railroad Ave Ft Morgan, CO 80701	Phone: 970-867-4918 x 257 Fax: 970-867-0878 Email: paulg@nchd.org	Logan, Morgan, Phillips Sedgwick, Washington, Yuma

Regional Office	*Discipline Contact and Address*	*Contact Information*	*County(ies)*
Northwest	Beth Watson Northwest Colorado Visiting Nurse Association, Inc. 940 Central Park Drive, Suite 101 Steamboat Springs, CO 80487-8816	Phone: 970-879-1632 x 7619 Fax: 970-870-1326 Email: ewatson@nwcovna.org	Grand, Jackson, Moffat Rio Blanco, Routt
Pueblo	Lynn Procell Pueblo City-County Health Department 151 Central Main Street Pueblo, CO 81003-4297	Phone: 719-583-4368 Fax: 719-583-4439 Email: lynn.procell@co.pueblo.co.us	Pueblo
South Central	Sue Foster South Central HCP Regional Office 1570 12th Street Alamosa, CO 81101	Phone: 719-589-4313 Fax: 719-589-2073 Email: suefoster@qwest.net	Alamosa, Chaffee Conejos, Costilla, Custer Fremont, Huerfano, Lake Las Animas, Mineral Rio Grande, Saguache
Southeast	Lorene Nelson Otero County Department of Health 811 South 13th Street Rocky Ford, CO 81067	Phone: 719-383-3047 Alt. Phone: 719-254-5300 Fax: 719-254-5310 Email: lnelson@oterogov.org	Baca, Bent, Cheyenne Crowley, Kiowa, Kit Carson, Lincoln, Otero, Prowers
Southwest	Liza Tregillus San Juan Basin Health Department PO Box 140 Durango, CO 81302	Phone: 970-247-5702 x 205 Fax: 970-247-9126 Email: liza@sjbhd.org	Archuleta, Dolores, La Plata Montezuma, San Juan
Tri-County Health Department - HCP	Jamie Gury Tri-County Health Department 4857 South Broadway Englewood, CO 80110	Phone: 303-783-7127 Fax: 303-761-1528 Email: jgury@tchd.org	Adams, Arapahoe, Douglas, Elbert
Weld	Lori McCarty Weld County Department of Public Health and Environment 1555 North 17th Avenue Greeley, CO 80631	Phone: 970-304-6420 x 2309 Fax: 970-304-6416 Email: lmccarty@co.weld.co.us	Weld
Western Slope	Kathie Talkington Mesa County Health Department PO Box 20000-5033 Grand Junction, CO 81502-5033	Phone: 970-248-6940 Fax: 970-248-6913 kathie.talkington@mesacounty.us	Delta, Eagle, Garfield Gunnison, Hinsdale Mesa, Montrose Ouray, Pitkin, San Miguel Summit
	Colleen Roahrig Mesa County Health Department PO Box 20000-5033 Grand Junction, CO 81502-5033	Phone: 970-248-6944 Fax: 970-248-6913 Email: colleen.roahrig@mesacounty.us	