Approved March 15, 1984
Date

MINUTES OF THE SENATE COMMITTEE ON PUBLIC HEALTH AND WELFARE.

The meeting was called to order by Senator Jan Meyers at
Chairperson

10 a.m./p.m. on March 13, 1984 in room 526-S of the Capitol.

All members were present except:

Senator Francisco, excused

Committee staff present:

Emalene Correll, Legislative Research Department
Bill Wolff, Legislative Research Department
Norman Furse, Revisor of Statutes office

Conferees appearing before the committee:

Rep. Homer Jarchow
Dr. Joseph Hollowell, Department of Health and Environment
Lynelle King, Kansas State Nurses Association
Joan Strickler, Executive Director. Kansas Advocacy and Protective Services for the Developmentally Disabled, Inc.
Ethel May Miller, Topeka

Others present: see attached list

HB 2864 - Requiring diagnostic tests on infants to determine if the child has galactosemia

Rep. Homer Jarchow testified in support of HB 2864, and distributed written testimony which included an article from the Saturday Evening Post entitled "A Mentally Retarded Baby Is Forever". Rep. Jarchow stated that this bill adds the requirement of screening and testing new born babies for galactosemia, and over 30 states now include this screening and testing. (Attachment #1). He also distributed to the committee a copy of the testimony of Dr. Virginia E. Tucker, Medical Director of the Crippled and Chronically Ill Children's Program, DH&E, when she testified in support of this bill before the House Public Health and Welfare Committee. In her testimony Dr. Tucker stated that DH&E recommends that this bill be passed favorably, provided adequate resources are identified to carry out the testing and the follow-up, and DH&E would prefer that this test become a requirement beginning January 1, 1985. (Attachment #2).

In answer to questions from the committee, Rep. Jarchow said that the fiscal note on this bill is approximately $41,000, and that at the present time he know of two cases of galactosemia in Kansas. There was also discussion as to setting an earlier effective date for this required testing.

Dr. Joseph Hollowell, DH&E, testified in support of HB 2864, and stated that this is a preventable genetic disease, if detected early enough. He distributed written testimony from DH&E supporting this bill. (Attach. #3).

In answer to a question as to why the delay until January, 1985, before this testing is required, Dr. Hollowell answered that there are a number of laboratories in the state, and they couldn't be assured that a standardized process could be set up before then. Dr. Hollowell said the cost to parents for the existing required test is between $10 and $15, and a third screening test would probably add a little to the cost. About a third of the tests are done in private laboratories, and if the state does the tests, they do not charge.

Lynelle King, KSNA, testified in support of HB 2864, and distributed testimony stating that KSNA unequivocally supports this bill. She also submitted a copy of the testimony presented by Evelyn Smith, RN, to the

House Public Health and Welfare Committee on this bill. Included in this testimony was an article written by Ms. Smith on "Galactosemia: An Inborn Error of Metabolism". (Attachment #4).

Joan Strickler, Executive Director, KAPS, testified in support of HB 2864, and distributed testimony stating that early diagnosis and treatment are imperative, and passage of this bill will make it possible for Kansas to make wise use of the medical knowledge we now have available about galactosemia. (Attachment #5).

Ethel May Miller, Topeka, testified in support of HB 2864, stating that the Kansas Association of Retarded Citizens has long been active in promoting prevention activities.

SB 190 - Certificates of registration as psychologists to be issued to certain persons holding a master's degree

Senator Johnston moved that SB 190 be reported adversely. Senator Morris seconded the motion and it carried.

HB 2501 - concerning nursing; affecting the qualification of applicants for licenses to practice

Senator Johnston moved that HB 2501 be reported adversely. Senator Morris seconded the motion and it carried.

SCR 1648 - concerning investigations and hearing by the Board of Embalming

Senator Meyers said that this bill corrects a typographical error, which omitted a sentence in the bill.

Senator Morris moved that SCR 1648 be reported favorably and placed on the Consent Calendar. Senator Gordon seconded the motion and it carried.

HB 2095 - relating to traffic control, SRS institutions

Senator Johnston moved that HB 2095 be reported favorably. Senator Ehrlich seconded the motion and it carried.

HB 2101 - Divorce and annulment statistics, reports to SRS

Senator Morris moved that the bill be amended as suggested by Norman Furse, Revisor of Statutes office. Senator Hayden seconded the motion and it carried.

Senator Ehrlich moved that HB 2101 be reported favorably, as amended. Senator Hayden seconded the motion and it carried.

HB 2996 - Temporary permits issued by Board of Healing Arts

Senator Morris moved that the bill be amended by inserting language that those already having temporary permits would not be affected. Senator Gordon seconded the motion and it carried.

Senator Johnston moved that HB 2996 be reported favorably, as amended. Senator Ehrlich seconded the motion and it carried.

MINUTES OF THE SENATE COMMITTEE ON PUBLIC HEALTH AND WELFARE,

room 526-S, Statehouse, at 10 a.m./p.m. on March 13, 1984.

HB 2094 - relating to rehabilitation and halfway house advisory committee

Senator Vidricksen moved that HB 2094 be reported adversely. Senator Ehrlich seconded the motion and it carried.

HB 2864 - Diagnostic tests on infants to determine if child has galactosemia

The committee discussed taking action on this bill, and there was some question whether the private laboratories would be ready if an earlier effective date for testing was set. Dr. Hollowell said he would bring in more information on this, and the committee delayed taking any action until hearing further from Dr. Hollowell.

Senator Morris moved that the minutes of February 27, 28, 29, 29 (Noon), and March 1, 1984, be approved. Senator Gordon seconded the motion and it carried.

The meeting was adjourned.

SENATE
PUBLIC HEALTH AND WELFARE COMMITTEE
DATE 3-13-84

(PLEASE PRINT)

NAME AND ADDRESS	ORGANIZATION
Homer E. Jarchow	St Repres 95th Dist
Joan Strickler	KAPS
Lynelle King	Ks State Nurses Ass
Julie Fry	observing KU Law School
[illegible]	[illegible]
Joe Hollowell	KDHE
Keith R Landis	CHRISTIAN SCIENCE COMMITTEE ON PUBLICATION FOR KANSAS
[illegible]	[illegible]
Bonnie J. Palmer	[illegible]
Bill Palmer	Iola, Newton, Ks.
[illegible]	[illegible] KS
[illegible]	[illegible] Ks
Elinor Fosenine	" " "
Marjorie Weil	" " "
Marian Bussey	" " "
Olga Busenitz	" " "
M. Hawver	[illegible]
Nancy Wood	PTA
Shirley Douglas	PTA

H1 - 3-13-84

STATE OF KANSAS

HOMER E. JARCHOW
REPRESENTATIVE, NINETY-FIFTH DISTRICT
SEDGWICK COUNTY
2121 WEST DOUGLAS
WICHITA, KANSAS 67213

TOPEKA

HOUSE OF
REPRESENTATIVES

COMMITTEE ASSIGNMENTS
MEMBER ASSESSMENT AND TAXATION
COMMERCIAL AND FINANCIAL
INSTITUTIONS

March 13, 1984

HOUSE BILL NO. 2864

I WOULD LIKE TO THANK THE CHAIRPERSON FOR THE HEARING ON HOUSE BILL 2864. MY DAUGHTER-IN-LAW ALSO THANKS YOU. IT WAS HER IDEA THAT I PURSUE THE ISSUE CONTAINED IN THE BILL.

HOUSE BILL NO. 2864 ADDS THE REQUIREMENT OF SCREENING AND TESTING FOR GALACTOSEMIA ON NEW BORN BABIES. AS YOU CAN TELL BY THE BILL PKU AND HYPOTHYROID SCREENING AND TESTS ARE PRESENTLY REQUIRED. THE TEST FOR GALACTOSEMIA CAN BE ACCOMODATED FROM THE SAME DROPLET OF BLOOD TAKEN FROM THE BABY THAT IS USED FOR THE PKU AND HYPOTHYROID TESTS. OVER 30 STATES NOW INCLUDE THIS SCREENING AND TESTING.

GALACTOSE IS A MILK SUGAR THAT NEEDS AN ENZYME TO CONVERT IT INTO GLUCOSE OTHERWISE THE GALACTOSE ACCUMULATES DANGEROUSLY IN THE BLOOD. THE CORRECTION IS TO TAKE THE BABY OFF MILK IMMEDIATELY OR, IDEALLY, NEVER BE STARTED ON MILK. IF MILK FEEDING IS CONTINUED, THE BABY WILL NOT ONLY BECOME MENTALLY RETARDED, BUT WILL DIE AT AN EARLY AGE.

DR. CHO, A PEDIATRICS AND GENETICS EXPERT WITH THE KU MEDICAL CENTER, WICHITA BRANCH APPEARED AS A PROPONENT BEFORE THE HOUSE COMMITTEE. DR. TUCKER FROM THE DIVISION OF HEALTH ALSO APPEARED AS A PROPONENT. BOTH REGRET THEY CANNOT BE HERE TODAY.

Atch. 1

IN ADDITION DRS. HOLLOWELL AND SCHLOESSER OF THE DIVISION OF HEALTH AND DRS. NEIL SCHIMKE AND LEONA THEROU OF THE KU MEDICAL CENTER HAVE ADVISED ME THEY SUPPORT THE LEGISLATION BUT BUSY SCHEDULES KEPT THEM FROM APPEARING.

THERE IS A FISCAL NOTE ON THE BILL. OF THE APPROXIMATE $41,000 EXPENDITURE $12,500 WILL BE FOR EQUIPMENT AND $5,000 FOR THE COST OF FOLLOW-UP EDUCATION ACTIVITIES AND APPEAR TO BE NON-RECURRING IN NATURE.

ARE THERE ANY QUESTIONS?

ATTACHMENTS: COPY OF THE SATURDAY EVENING POST ARTICLE.

Speaking Out

A MENTALLY RETARDED BABY IS FOREVER

Severe mental retardation and death from galactosemia can be prevented with screening and early treatment. If mothers passed the laws, all babies would be screened. You can help.

by Cory SerVaas, M.D.

State legislatures are important because they can pass laws that change people's lives.

I can't think of a more drastic change in a young woman's life than to become the mother of a mentally retarded child, when for a few cents a day, that child, if treated in the first days or weeks of life, could have developed a normal brain.

All but a few of our states have passed laws that require *all* babies to have a heel prick with a droplet of blood drawn to test whether the child may have hypothyroidism.

One out of 4,000 infants born in the United States is found to have hypothyroidism. For a pittance, the infant is given a readily available thyroid hormone that will allow its brain to grow normally. It is estimated that nearly 100 percent of hypothyroid infants treated at one month will have normal mental development, compared to 80 percent treated at three months and less than 40 percent after four months. Several studies have shown that roughly 50 percent of brain growth occurs in the first six months of life. Untreated hypothyroidism in this time period has disastrous complications in the baby's brain.

Yet there are no symptoms, and there is nothing about the pregnancy of the healthy mother who bears a hypothyroid baby to give even a clue that the baby will not develop normally. No doctor can tell by physical examination. The most experienced pediatrician cannot tell.

At birth, this baby does everything that a normal, healthy baby is supposed to do. There are no symptoms—not a single sign.

But in the laboratory the facts contained in the baby's blood are swiftly apparent. The blood shows that the thyroid hormone is missing. The blood sample can be a little, dried spot of blood smaller than your fingerprint. Dried blood can be transported across the country on blotter paper. It doesn't have to be kept fresh. But that sample can change the course of a child's life and certainly the life of the child's mother as well.

Obviously, no infant in this

We asked Dr. Richard Schreiner (left), a neonatologist at Indiana University, to demonstrate the heel prick on baby Seigel. A number of tests to prevent mental retardation can be done on a very small droplet of blood.

civi.. country should be permitted to grow up mentally retarded from this highly preventable cause. A small amount of thyroid hormone replacement given orally every day allows the infant's brain to grow and function normally.

The only remaining states that do not have the mandatory testing law are Virginia, Mississippi, Nebraska and Hawaii. These states have voluntary programs for screening their newborns, but they are not mandatory. Virginia has passed a law requiring hypothyroid screening that will be in effect on July 1, 1984.

Another preventable cause of mental retardation is phenylketonuria (PKU). PKU is the inability of the infant to metabolize the amino acid phenyalanine, an amino acid present in protein that accumulates in the blood. By decreasing this amino acid in the diet during the infant's rapid-brain-growth period, mental retardation can be avoided. Testing for PKU is mandatory in every state except Mississippi.

Galactosemia

The test that The Saturday Evening Post Society wants to crusade for at this time is another enzyme deficiency test that can be easily detected—from the same drop of blood already mandatorily drawn from the baby for the PKU and hypothyroid tests. The next most common cause of preventable mental retardation is *galactosemia*, which simply means galactose in the blood. Galactose is a milk sugar that needs an enzyme to convert it into glucose; otherwise the galactose accumulates dangerously in the blood. If the baby doesn't have the enzyme, it will be fine as long as it isn't fed galactose. The only natural source of galactose is primarily in dairy products, so the baby should be taken off milk immediately or, ideally, never be started on milk. If milk feeding is continued, the baby will not only become mentally retarded, but will die at an early age.

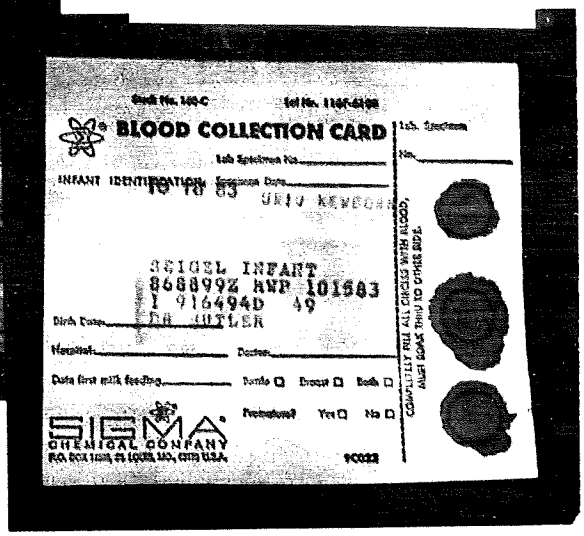
In one typical family the first child, a boy, died from what was presumed to be pneumonia at one month of age. Unfortunately, at the time their second child, a girl, was born, her state, California, did not yet require routine screening for galactosemia. The baby failed to gain weight and was sent for extensive testing. Galactosemia was finally diagnosed when she was one month old. The disease process was arrested then as all dairy products were removed from her diet, but damage from one month's lost time left her with cirrhosis (hardening) of the liver and cataracts, along with some learning disabilities. At ten she is having difficulty learning to read. Her mother hopes screening will soon be mandatory in all states. She says, "I don't think a parent should have to go through the loss of a child in order to save the next one."

"One third to one half of the families that have a galactosemic child eventually diagnosed have already had one infant die with a disease that, in retrospect, at autopsy or clinically, looked like galactosemia," said Dr. Richard Koch, Professor of Clinical Pediatrics at the U.S.C. School of Medicine and Head of the Division of Medical Genetics at the Children's Hospital of Los Angeles. I asked him how many galactosemic patients were being discovered. "We're running about one in 40,000 here in California," he replied. "It is slightly more common in Dutch and Northern European groups. Galactosemia occurs in blacks at about the average incidence." In discussing the cost, Dr. Koch said, "All you have to do is find one patient, and it pays for all the screening."

The best way to get a law passed requiring that all newborns be tested for galactosemia in your state is to write to your own state representative and state senator asking them to introduce a bill.

In California the incidence of galactosemia has been about one in 40,000. If your state has 400,000 births a year and it doesn't screen for galactosemia, you could be sure of saving ten children from mental retardation and death—if you could get the mandatory testing law put into effect.

You can make a difference.



The blotter paper (right), containing baby Seigel's blood, will be sent to a laboratory to rule out the possibility of PKU and hypothyroidism. The same sample *could* be used to test for a galactose defect. In California, Ohio, Texas and Massachusetts it would be. What about your state?

#2- 3-13-84
Attm 4
2-29-83

KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

TESTIMONY ON HOUSE BILL NO. 2864

FEBRUARY 29, 1984

HOUSE PUBLIC HEALTH AND WELFARE COMMITTEE

This is the official position taken by the Kansas Department of Health and Environment on House Bill 2864:

House Bill 2864 adds the screening for galactosemia to the newborn screening law first enacted in 1965 to screen for phenylketonuria and amended in 1977 to add congenital hypothyroidism. The law requires the department to conduct educational programs, provide recognized screening, diagnostic and treatment control tests, maintain a registry of cases, and provide the necessary treatment product for diagnosed cases of phenylketonuria, hypothyroidism, and such other diseases as may be appropriately detected with the same procedures for which laboratory services are required.

Infants born with galactosemia have an intolerance to the sugars, lactose and galactose, as the result of one of two enzyme deficiencies, galactose - 1 - phosphate uridyltransferase, or galactokinase. The transferase deficiency is the most common and the most severe, ofter resulting in early death of the infant. Symptoms usually do not appear until the infant starts milk feedings since the principal sugar in milk is lactose. The abnormality is manifest in organ systems by the following signs and symptoms: the eye - lenticular catarracts; the liver - hepatomegaly, jaundice, hypoglycemia; the kidney - generalized aminoaciduria; the gastrointestinal tract - anorexia, vomiting, abdominal distention, ascites, cholelithiasis, acholic stools; the central nervous system - lethargy, hypotonia, mental retardation. Mental retardation appears to be irreversible when it occurs.

Treatment of the disease is restriction of foods containing lactose and galactose from the patient's diet the remainder of life in the majority of cases. With this treatment measure, largely the replacement of milk and milk products with other nonlactose containing nutrients, the child can develop normally and lead a normal life. Otherwise the outcome can be devastating to the child and family.

The disorder is transferred as an autosomal recessive trait. The prevalence of the disorder is 1:50,000.

Over 30 states have added the test for galactosemia to the basic screening tests for phenylketonuria and hypothyroidism and a number of states have pilot programs to evaluate the usefulness of adding this test to the general screening program. In Kansas by adding this screening procedure the Kansas Department of Health and Environment laboratory would require additional staffing and funds for equipment and supplies. Approximately 1/3 of newborns in Kansas have genetic screening performed by eight hospital laboratories and one independent laboratory rather than the State Health Department laboratory. A recent survey of these hospitals indicated that they would cooperate in testing for galactosemia if it became a required procedure for routine screening.

Atch. 2

There would be a problem for the Kansas Department of Health and Environment laboratory as well as private laboratories to get the test started up and on-line before January 1, 1985. It would be preferred that should this test become a requirement for newborns in Kansas that the requirement begin January 1, 1985. There would be initial start up costs for equipment and supplies and an ongoing cost of laboratory personnel and follow-up in educational activities which would continue after the first year.

The Department of Health and Environment recommends that this bill be recommended favorably for passage provided adequate resources are identified to carryout the testing and the follow-up.

Presented by: Virginia E. Tucker, M.D., Medical Director
Crippled and Chronically Ill Children's Program
Kansas Department of Health and Environment

A 3 - 3-13-84

KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

TESTIMONY ON HOUSE BILL NO. 2864

PRESENTED MARCH 13, 1984

SENATE PUBLIC HEALTH AND WELFARE COMMITTEE

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The problem expressed by the Kansas Department of Health and Environment laboratory as well as private labs to get the tests started and on-line before January 1, 1985 has been corrected with the House amendment so that it will become a requirement for newborns in Kansas on January 1, 1985. There would be initial start up costs for equipment and supplies and an ongoing cost of laboratory personnel and follow-up in educational activities which would continue after the first year.

Att. 3

The Department of Health and Environment recommends that this bill be recommended favorably for passage provided adequate resources are identified to carry out the testing and the follow-up.

Presented by: Joseph G. Hollowell, Jr., M.D., Director
Division of Health
Kansas Department of Health and Environment

#4 3-13-84

KSNA
the voice of Nursing in Kansas

Statement of the Kansas State Nurses' Association
by Lynelle King, R.N., M.S., Executive Director
before the Senate Public Health and Welfare Committee
March 13, 1984

In support of HB 2864 - Tests for Galactosemia

KSNA unequivocally supports this bill. One can, while doing the other tests already required for newborns, also test for Galactosemia and thus make it possible to prevent mental retardation which would otherwise result from this hereditary condition. Not to require this test is unconscionable when one realizes the consequences.

Attached is a copy of our more thorough testimony given on the House side by a nurse who has studied infants with galactosemia.

Atch. 4

KSNA
the voice of Nursing in Kansas

Statement of Kansas State Nurses' Association by Evelyn Smith, R.N. before the House Public Health & Welfare Committee
February 29, 1984
Supporting HB 2864 Requiring Diagnostic Test on Infants to Determine If They Have Galactosemia

Mr. Chairman and members of the committee, my name is Evelyn Smith. I am a family nurse practitioner as well as an educator at Wichita State University. I represent the Kansas State Nurses Association. Galactosemia, though rare, is a diagnosable genetic disease in newborns. Because time is of essence and milk creates problems for these babies, screening may be a lifesaving procedure. I have written an article which includes a case study of such an infant. With proper diagnosis and elimination of milk products those infants may live a fairly normal life.

There are 30 states already routinely screening babies for galactosemia as of April 1983. The cost of screening may range considerably depending on whether it is done on a single basis or multiple screening. From the Infant Screening article, you will note that there are government grants available which encourage such screening.

As I stated in 1980 routine screening is necessary to diagnose galactosemia in the newborn infant before the infant becomes ill. From the research I have done I would personally support such screening in the state of Kansas.

GALACTOSEMIA: AN INBORN ERROR OF METABOLISM

Evelyn J. Smith, R.N.,C., B.S.N.

As a clinical entity, galactosemia was first described in 1908. Not until 1956 was it defined as resulting from a specific enzyme defect. We now know enough about it to describe its mechanism of action, as well as how its structure and function are modified by its various structural gene mutations. "Galactosemia is an inborn error of carbohydrate metabolism in which the body is unable to utilize the sugars galactose and lactose."[1] The reason for the inability to utilize the sugars is an absence of the enzyme galactose 1- phosphate uridyl transferase in the liver.

Normally in liver cells appropriate enzymes are available to promote interconversions between the monosaccharides. The final product is glucose, the sugar that can be utilized by the cells of the body for energy. In galactosemia, one step in that conversion process is absent due to a gene mutation which is responsible for the absence of galactose 1- phosphate uridyl transferase (Gal 1-PUT). As a result galactose circulates unusable in the blood and is excreted in the urine. Cataracts, brain damage, and hepatomegaly with fatty metamorphosis in fatal cases occur.

In an autosomal recessive inheritance pattern the parents must be heterozygous for the trait and usually do not display any symptoms; they are carriers. Males and females are affected with equal frequency. One fourth of the children of parents who are unaffected will be affected. A negative antecedent family history is the usual pattern.

Symptomalogy

Infants appear normal at birth but within a few days they begin to vomit and lose weight. Jaundice and liver enlargement are also early signs. The baby soon appears sleepy, exhibiting nausea, vomiting and diarrhea. Septisemia appears in a few days. Clinical symptoms may lead to severe mental retardation and cataracts, as well as cirrhosis of the liver, if the infant survives but remains untreated.

Diagnosis

The primary health care provider must rule out neonatal jaundice, kernicterus and hypoglycemia when confronted with an enlarging liver. Then studies of galactose 1- phosphate uridyl in RBCs and laboratory levels of galactose in the blood stream or urine should be obtained. Levels of galactose in the urine may not be elevated right away so one must not depend on one lab test alone or discontinue pursuit of the problem when dealing with a sick infant.

Case History

A boy was delivered at term, weighing 3.4 kg, after an uneventful pregnancy. He was the second child in the family. The first child had failed to thrive and died at the age of one month from a generalized fulminating septisemia. The second baby was well until day six, when he fed poorly, failed to gain weight and was found to have a *Klebsiella* septisemia. He improved, over the next week on antibiotics. During this time, however, he became jaundiced with a maximum serum bilirubin (5.85 mg/100 ml) and his liver became palpable, 2 cm below the right costal margin. He had recurrent thrush infections and a low serum IgA. At five weeks, he developed diarrhea; stool cultures yielded *Alkalescens dispar* but other cultures were negative and he improved once again on antibiotics. His hepatomegaly was 3 cm below the right costal margin at six weeks, but he was reported to be feeding well on Ostermilk®. A urine report at that time was negative for reducing substances. At eight weeks he was said to be feeding well and gaining weight, but a Benedicts' test for urine reducing substances was again negative. At nine weeks he was well, but hepatomegaly persisted. Urine was tested again, and showed a trace of galactose with amino-aciduria. A red cell assay for Gal 1-PUT, showed no enzyme activity. He was started on a galactose free diet and progressed well. He is now two years old and making normal developmental progress with no evidence of cataracts.[2]

This child has classic galactosemia. The diagnosis was missed until he was twelve weeks old. The limitations of urine screening tests are obvious. A wider use of qualitative enzyme assay needs to be utilized.

Diagnosis

The urine test may continue to be the basic screening test for galactosemia, but is obviously of no value in an infant who is not feeding regularly on milk. In a sick infant a negative result for reducing substances should not be used alone to exclude galactosemia, in the face of clinical symptoms. In all cases of doubt, an enzyme screening test should be performed.

Beutler and Baluda devised a screening method in 1966, using filter paper with blood spots similar to PKU testing. Further screening for Gal 1-PUT requires ultra-violet light to show the presence of the enzyme in red cells. With classic galactosemia the red cells produce no fluoresence.

Routine newborn screening is necessary for the diagnosis of galactosemia. The full impact of galactosemia of the newborn infant has become evident only since routine screening has been initiated in some states. It is now apparent that death associated with bacterial sepsis may occur in about 30 percent of those with untreated classic galactosemia. This usually occured by the second week of

life. procedures are: urine should be tested for redu tances; if urinary reducing substance is found, osemia should be presumed. If urinary reducing substance is found, milk feedings should be discon-inued and blood and urine specimens sent to the lab for confirmatory testing. If the infant is ill, bacterial cultures should be obtained and treatment for sepsis initiated. If galactosemia is confirmed treatment should be continued.

Treatment and Counseling

When positive results are found, patients and their families need to be investigated further by a quantitative assay for Gal 1-PUT, so that counseling can be initiated. Then, if prenatal diagnosis is considered, a precise knowledge of familial genotypes is available.

Any one of several phenotypes of galactosemia is likely to be found in the infant with an abnormal screening test. Phenotyping is important as the genetic form of galactosemia may determine the prognosis, course of treatment, and need for genetic counseling.

Treatment of the infant includes galactose free formula, called Nutramigen®, or meat-base formula. Nutramigen is a protein hydrolysate process formula produced by Mead-Johnson. The various formula companies usually have their own brand of meat-base formula. The nurse, in counseling parents about their baby's condition, should suggest several, and know the most economical way of purchasing, as this special formula is more costly than regular formula. If the clinician presents an economical route of purchase, compliance may be higher. As baby grows, parents must be educated, not only to avoid giving their child milk, but to carefully omit any foods containing milk. Some foods have dry skim milk added. Among those are most breads, crackers, pastries and cakes. Butter, margarine and cheese should also be avoided. Brain tissue contains galactose and must be omitted. Labeling must be checked and if a product is not clear, it should be avoided. Parents may have to change their food preparation and eating style somewhat to incorporate their child's needs into their own diet. Processed, quick foods may not be feasible for such families. A good nutritious variety of food can be offered with ood planning. The nurse can fill this role of educating arents by taking a family nutritional history and giving lists and suggestions of how to adapt to their child's diet. Parents must be cognizant of the consequences if they fail to follow the prescribed diet. They must understand that this child will *never* be able to tolerate milk or milk products. Depending on a diet without milk, calcium and vitamins need to be calculated on the child's needs, especially vitamins A and D.

As soon as the parents have adjusted to the shock and crisis of having a "different" child, they will become concerned about having more children with the same problem. As discussed before, galactosemia is an autosomal recessive disease and parents have a 25 percent chance of having another child with galactosemia and that 50 percent of the children will be carriers. Other alternatives to having their own children should be offered. Prenatal diagnosis is an option for these parents, and should be discussed. Include where it can be done, how much it costs, risks involved, and what is usually expected of them if a positive report is received. Justification of amniocentesis may arise, since these children can be treated. The procedure can remove uncertainty in the minds of parents. Further, knowledge that an affected fetus is to be delivered insures immediate, proper treatment.

Even with prompt diagnosis and treatment of the disease, these children may still be undersized and of low intelligence.[4] They may have coordination and perceptual disabilities which are thought to be associated with their low intelligence. One study indicated such children had more emotional disturbances and were particularly sensitive to criticism. They tended to have poor relationships with the adult world.[4] Parents will continue to feel frustrated by their child's problems. Referral may be needed at some time for family counseling. The nurse can help parents set realistic goals for their child on the basis of individual testing of capabilities, usually done through the public schools. Assessment of vision and fundoscopic examinations to rule out cataracts are essential in follow up care.

As the child enters adolescence and even before, he must also be educated about his or her condition to prevent noncompliance of diet. This is a common problem in pediatrics. Children at this stage do not want to be different. As the child grows into adulthood, further genetic counseling needs to be done regarding marriage and children. (NP)

REFERENCES

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To enhance the development of babies...
INFANT SCREENING
Volume 5, No. 1, April, 1983

PROFILES IN PREVENTIVE MEDICINE

Editor's Note

Good medicine cures, great medicine prevents disease. Yet the pioneers who developed preventive medicine in the field of infant screening fought a hard battle against medical forces still wedded to the treatment-for-service and cost-effective approaches in pediatrics. Profiles of these farsighted pioneers will be presented in the newsletter from time to time. We are pleased to initiate the series with a profile of a great innovator, Dr. Robert Guthrie, who launched the program of infant screening in medicine.

Dr. Robert Guthrie

"If I have made any significant contribution to science and human betterment, it was my development of screening tests to detect rare metabolic conditions in infants, such as phenylketonuria (PKU)." (Blatt, in press) Thus, Dr. Robert Guthrie begins a chapter about himself and his PKU test in a book about the lives of people who contributed to the field of mental retardation. His chapter is entitled "Explorations in Prevention".

After growing up in a small town in Marianville, Missouri, Guthrie went to the University of Minnesota and earned a bachelors degree, a Ph.D. and an M.D. He lso holds a masters degree from the University of Maine. He never practiced medicine, though, choosing instead to be a medical researcher.

In 1946, Guthrie began working at the National Institutes of Health as the only scientist working there in biochemical genetics, a new field at the time. In 1951, Guthrie moved his family to Staten Island and began working at the Sloan Kettering Institute for Cancer Research in New York City on problems related to childhood leukemia. While living on Staten Island, his six year old son was diagnosed as being mentally retarded.

In 1954, Guthrie moved to Buffalo, New York, to join the staff of the Roswell Park Memorial Institute, to continue his cancer research, specializing in microbial biochemical genetics. Buffalo's Children's Hospital asked Guthrie to develop a method of measuring blood phenylalanine to help with the newly developed treatment for phenylketonuria. He applied similar methods he was already using in biochemical genetics to quickly develop a bacterial test to measure blood phenylalanine quantitatively. This was done by combining bacillus subtilis in a simple agar culture medium with an available chemical antagonist of phenylalanine. By placing paper disks that had been punched from a filter paper impregnated with a measured amount of blood serum, he could estimate the amount of phenylalanine by comparing the diameter of the growth zones. The test was easy to perform since it only required a tiny amount of blood from the child.

Dr. Guthrie then moved over to the Department of Pediatrics at Children's Hospital and began studying the different microbial inhibitors available that had been synthesized for use in cancer chemotherapy. He hoped he could use this approach to find ways of detecting new metabolic diseases in children.

After he started this program in 1958, an amazing coincidence occurred. Guthrie learned that his wife's niece had just been diagnosed as having phenylketonuria. However, because she was not diagnosed until she was 14 months old, she had already become severely retarded.

At this point, Guthrie conceived the concept of applying the tests they already were using to monitor blood phenylalanine specimens from older children to newborn babies. If every baby could be tested, then the children who had phenylketonuria could be detected early and in time for treatment to prevent the otherwise almost inevitable mental deterioration. As Guthrie writes, "as it turned out, the test was even better than I would have dared to dream. The amount of blood on each one fourth inch (7 mm.) disk, punched with a paper punch from drops of blood spotted on filter paper, varies no more than approximately 3 to 5% in dry weight from one disk to another. This meant that an adequately quantitative sample could easily be obtained, similar to the liquid blood measured in a micro pipette in the usual way...Curiously enough, I have discovered as the years have gone by that no one, as far as I have been able to determine, in the entire history of medicine, has used dried spots of blood on filter paper, punched to obtain a quantitative sample of testing for any purpose."

Continued on page 2

Scre...ng Status Report

The status of screening programs across the country is constantly in the process of change. This is a state-by-state report, utilizing the most current information available. (Black dots are added to the list below a specific test only if every baby in a particular state is being screened for that particular disease. We would appreciate information from the individual state informing us when it is necessary to add or subtract a dot.)

	PKU	T-4	MSUD	Homocyst.	Galactosemia	Tyrosinemia	Sickle Cell	Other
Alabama	●	●					●	
Alaska	●	●	●	●	●	●		
Arizona	●	●	●	●	●		●	
Arkansas	●	●					●	
California	●	●			●		●	
Colorado	●	●	●	●	●		●	
Connecticut	●	●	●	●	●	●		
Delaware	●	●	●	●	●			
D.C.	●	●	●	●	●			
Florida	●	●	●		●			
Georgia	●	●	●	●	●	●	●	●[1]
Hawaii								
Idaho	●	●	●	●	●	●		
Illinois	●	●						
Indiana	●	●						
Iowa	●	●	●		●			
Kansas	●	●						
Kentucky	●	●			●			
Louisiana	●	●						
Maine	●	●	●	●	●			
Maryland	●	●	●	●	●			
Massachusetts	●	●	●	●	●			
Michigan	●	●						
Minnesota	●	●			●			
Mississippi								
Missouri	●	●						
Montana	●	●	●	●	●	●		
Nebraska	●							
Nevada	●	●	●	●	●	●		
New Hampshire	●	●	●	●	●			
New Jersey	●	●						
New Mexico	●	●	●	●	●		●	
New York	●	●	●	●	●		●	●[1] ●[3]
North Carolina	●	●						
North Dakota	●	●						
Ohio	●	●		●	●			
Oklahoma	●	●						
Oregon	●	●	●	●	●	●		
Pennsylvania	●	●						
Rhode Island	●	●	●	●	●			
South Carolina	●	●						
South Dakota	●	●						
Tennessee	●	●						
Texas	●	●		●	●			
Utah	●	●			●			
Vermont	●	●			●			
Virginia	●	●						
Washington	●	●						
West Virginia	●	●			●			●[1] ●[4]
Wisconsin	●	●	●		●			
Wyoming	●	●	●	●	●		●	

[1](on request)
[2]Hemoglubinopathies
[3]Adenosine-deaminase
[4]Histidinemia
(Alabama, Tennessee / at conference update)

Continued from page 1

The rest is history. The first pilot study began the summer of 1961 and, as a result of the screening, 23 cases of phenylketonuria were discovered at Newark state school. Actually, the first non-selective infant screening was started in a small hospital in Jamestown, New York. Then with funds from the Children's Bureau, Dr. Guthrie and his team rented a house close to Children's Hospital and converted it into a miniature factory to prepare enough test kits so that screening could start in 29 states. They prepared materials for a million tests. They set up quality control procedures to insure the materials were uniform. In the first countrywide testing of 400,000 babies who were screened, 39 cases diagnosed as PKU were detected.

Guthrie reports "Many of the doctors in the United States were not interested at all in the test. In fact, they were often very antagonistic to laws requiring the test. In all states except Massachusetts, state medical societies resisted the idea of a law, if they acted on the issue at all. Much of this attitude is related to the way in which our health care is organized. Much of the controversy concerning PKU screening and treatment was stimulated by resentment against the laws that were passed. Laws requiring babies to be tested were considered by many doctors as interference, an invasion by the government in a private medical matter. The rights of doctors seemed to be more important to some than the rights of infants to be protected from mental retardation."

Today, the battle for phenylketonuria appears to have been won in the United States (see Screening Status Report, on left) and also in many countries around the world. In the years since PKU testing was developed, 30 new tests have been devised in Guthrie's laboratory. Many of these tests are now used in newborn screening programs in areas of the United States and the world.

But Guthrie is not resting on his laurels. He is currently working towards a law to prevent children from becoming mentally retarded because of lead poisoning. In addition to his other duties for the past 12 years, he has been an integral part of a national team, pushing people to realize that lead poisoning is a major health problem in the United States. In 1980, he began testing lead specimens from children in the New York State area and found four children with lead levels so high they had to be hospitalized.

Thus, this indefatigable man is now lobbying against an environmental hazard, continuing his commitment to the most helpless members of our society—infants and young children.

Throughout the years since Dr. Robert Guthrie began promoting screening of infants for metabolic errors, he and his associates at the State University of New York at Buffalo have been most helpful to screening programs by providing assistance with quality control and procedure development. A grant from the U.S. Maternal and Child Health Service was renewed specifically to encourage and assist screening laboratories to expand their programs beyond PKU and hypothyroidism to include galactosemia, maple syrup urine disease, homocystinuria, and sickle cell disease.

For further information call 716-831-2351 or write to Dr. Guthrie at Acheson Hall, Room 352, 3435 Main Street, Buffalo, N.Y. 14214. Drs. Guthrie, Edwin Naylor, David Jinks and Mrs. Sally Bloom are willing to help with any problems. ■

#5 3-13-84

Kansas Advocacy & Protective Services for the Developmentally Disabled, Inc.

kaps

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To; The Senate Committee on Public Health and Welfare
Senator Jan Meyers, Chairperson

From: Kansas Advocacy and Protective Services
R.C. Loux, Chairperson

Date: March 13, 1984

Re: H.B. 2864

Galactosemia is a metabolic inability to convert galactose to glucose. Left untreated, infants may die. Survivors of this condition can be mentally retarded and have other serious impairments.

Early diagnosis and treatment is imperative. The prognosis is good if galactose is eliminated before birth and until age 6.

Passage of H.B. 2864 will make is possible for Kansas to make wise use of the medical knowledge we now have available about galactosemia. Through effective use of education, screening, diagnosis and treatment we can prevent serious handicapping conditions from occuring in the future.

Respectfully submitted,

Joan Strickler
Executive Director

JS/jw

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