



“ These kids are just like other children all over the world and they deserve a chance to live the best lives possible. ”

Clinical Geneticist Dr. Eugene Hoyme first became interested in FAS during his residency at the University of California. The effort to understand the disorder, improve diagnostic capabilities, and reduce its prevalence worldwide have become a career-long passion.

Eugene Hoyme, MD, and Fetal Alcohol Syndrome

By Alex Strauss

Sanford Researcher has Devoted Career to Understanding and Preventing FAS and Related Disorders

Fetal Alcohol Syndrome (FAS) and the related set of disorders that fall under the title Fetal Alcohol Spectrum Disorders (FASD) present many challenges for physicians, educators and researchers, not the least of which is the challenge of pinpointing their prevalence. CDC studies put the rate of FAS in the U.S. between 0.2 and 1.5 cases for every 1000 live births. Other studies using different methods have found it to be even more common – as many as 1 case out of every 500 live births. And scientists believe that there are at least three times as many cases of FASD as there are cases of FAS.

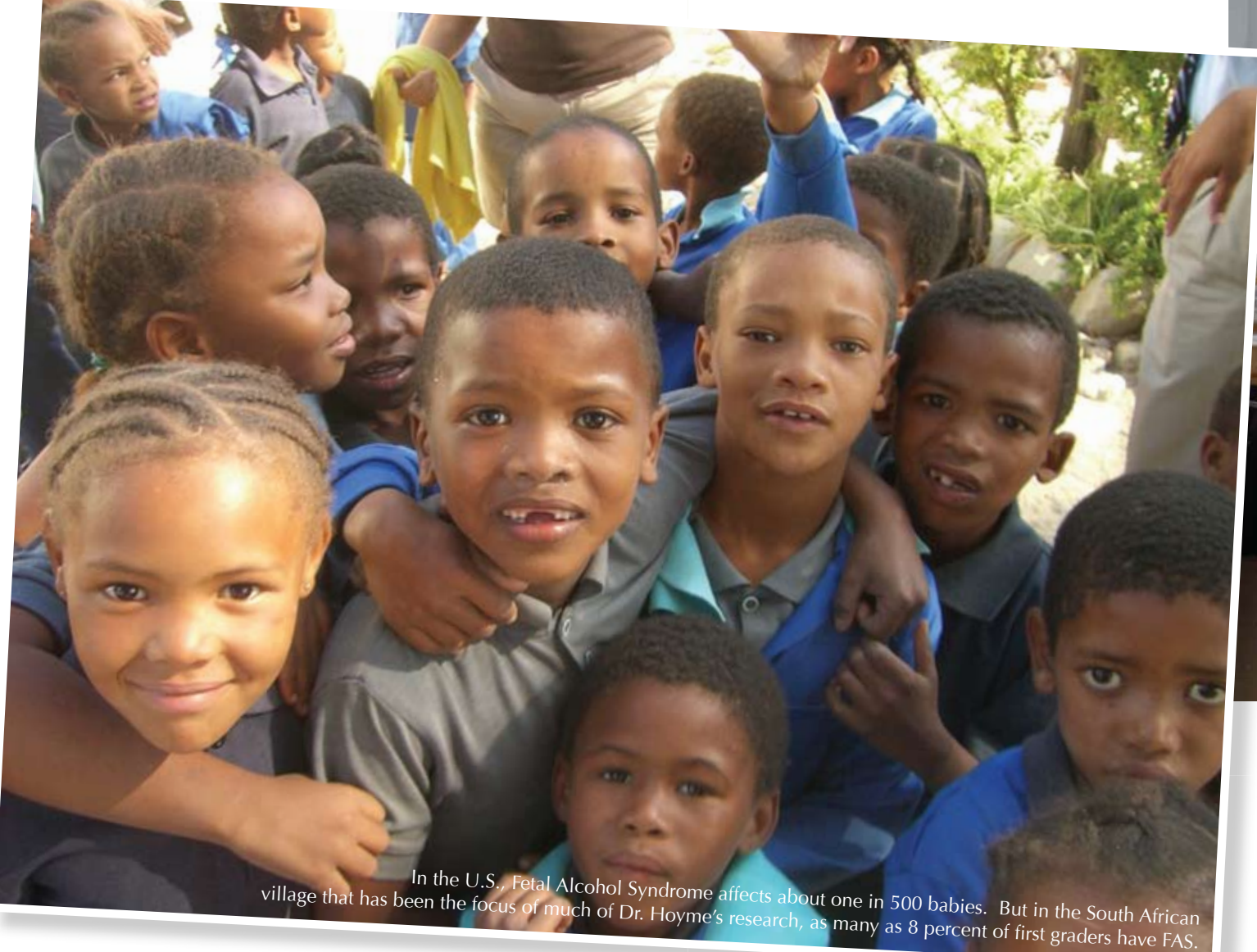
“There have only been two prevalence studies on FASD and both of them showed a rate of about one percent among school-aged children, across the entire spectrum of the disorder,” says Eugene Hoyme, MD, Professor and Chair of the Department of Pediatrics at USD’s Sanford School of Medicine.

Few physicians have seen as many cases of Fetal Alcohol Spectrum Disorders as Dr. Hoyme. He is the Chief Medical Officer of Sanford Children’s Hospital, Senior VP for Children’s Services at Sanford Clinic in Sioux Falls, and a renowned clinical researcher into the mysteries of these sometimes baffling disorders.

A graduate of Augustana College and the University of Chicago Pritzker School of Medicine, Dr. Hoyme’s fascination with FAS began more than 30 years ago during his pediatric residency at the University of California, San Diego.

“During my residency, the Chief of Pediatric Genetics there was Dr. Kenneth Jones, one of the first people to describe Fetal Alcohol Syndrome back in 1973,” recalls Dr. Hoyme. “Dr. Jones inspired many of us to go into the field of birth defects evaluation and care. He is also the one who invited me to stay on as a post doctoral fellow and to stay on in clinical genetics with him.”

Scientists estimate there are three times as many cases of FASDs as there are cases of FAS



In the U.S., Fetal Alcohol Syndrome affects about one in 500 babies. But in the South African village that has been the focus of much of Dr. Hoyme's research, as many as 8 percent of first graders have FAS.

Following his fellowship training in clinical genetics at the University of California, Dr. Hoyme went on to become a Professor and Chief of the Division of Medical Genetics and Associate Chair of the Department of Pediatrics at Stanford School of Medicine in Palo Alto, California, where he continued to expand and refine his study of FASD. He came to Sioux Falls and took on his post at Sanford and the School of Medicine in 2007. Dr. Hoyme is board certified in Pediatrics and Medical Genetics and has authored nearly 300 original articles, monographs, book chapters, research abstracts and a textbook. He has served on the editorial boards of five professional journals, including Pediatrics and Clinical Genetics.

Global Research Efforts

FASD was originally described by Drs. Kenneth Jones and David Smith in the early 1970's, although knowledge of the adverse effects of drinking during pregnancy had existed for centuries. These investigators were the first to define the fact that children of drinking mothers tended to have growth problems, both before and after birth.

They tended to be small children and small adults with smaller head circumference and restricted brain growth. Because FAS affects the development of the frontal part of the growing brain, the face tends to develop abnormally, resulting in tell-tale facial dysmorphism. According to the CDC, the following facial features are essential for a diagnosis of FAS: a smooth philtrum (no groove between the nose and upper lip), a thin vermilion border (thin upper lip), and small palpebral fissures (small eyelid openings, when adjusted for age and race).

“Most of these babies are not born to women who drink daily.... Most are born to women who are binge drinkers.”

A patient must meet all three of these criteria (growth deficiency, small head circumference and typical facial dysmorphism) to be diagnosed with FAS, although many other physical characteristics



As one of four clinical geneticists that make up the NIAAA's dysmorphology core, Dr. Hoyme has personally examined more than 3000 children who have been exposed to alcohol *in utero* and nearly 400 with full-blown FAS in the last 15 years.



Dr. Hoyme and a pair of twins in South Africa. To be classified as FAS, a patient must have growth deficiency, small head circumference and typical facial dysmorphism.

are often present and may include, among other things, malformed ears, a short nose, congenital heart defects, decreased mobility of the elbow joints, inability to completely extend the fingers, and small finger and toenails. Typically, prenatal or postnatal height and weight tend to be below the tenth percentile. FAS may also cause mild to moderate mental retardation, neurological deficit, mental disorders and developmental disabilities.

Although there is still much that is not fully understood about the prenatal effects of alcohol use, much of what is known has been uncovered by Dr. Hoyme and the other three clinical geneticists who form the dysmorphology core of the National Institute on Alcohol Abuse and Alcoholism (NIAAA) of the NIH. He and his colleagues have helped to more clearly define FAS and related disorders through years of research around the world.

“One of the most interesting things we have come to understand is that most of these babies are not born to women who drink daily,” says Dr. Hoyme. In the course of his research, Dr. Hoyme has personally examined more than 3000 children who have been exposed to alcohol *in utero* and nearly 400 with full-blown FAS in the last 15 years. “Most children with FAS are born to mothers who are binge drinkers, that is, they don’t drink during the week, but on the weekend may consume a case of beer, for example. That is a huge amount of alcohol for a fetus to be exposed to.”

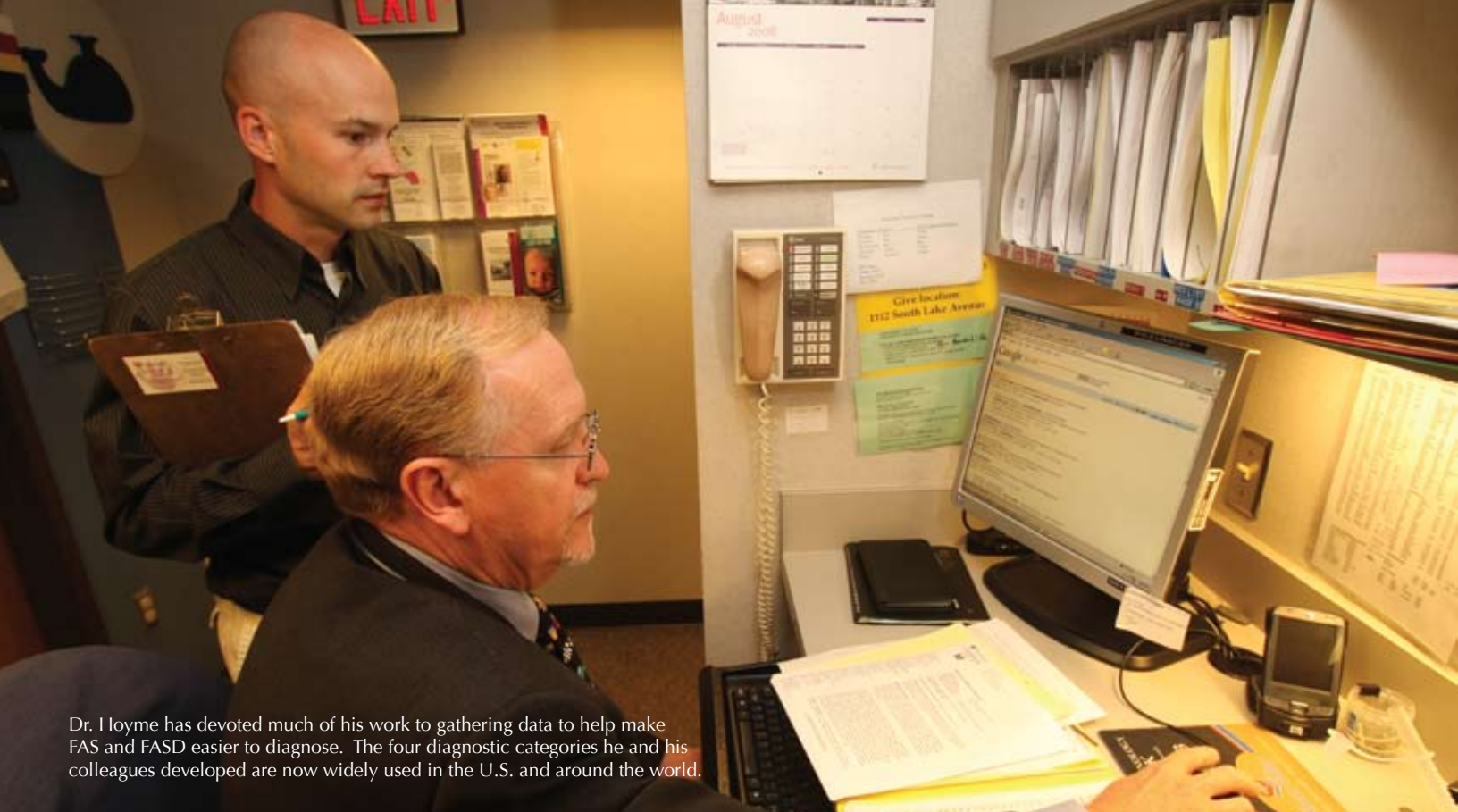
Dr. Hoyme’s current NIAAA research studies include projects through the “Collaborative Initiative on Fetal Alcohol Spectrum

Disorders” with his former colleague Dr. Kenneth Jones of the University of California, and “Fetal Alcohol Syndrome Epidemiology Prevention and Research” with Philip A. May, PhD of the University of New Mexico. Over the past 12 years, Dr. Hoyme’s research work has taken him to Africa for several weeks each year to study school children in a wine-growing region plagued by FASD.

“Our goal is to make this a condition that any physician can diagnose if he or she is careful and follows the guidelines.”

“For years our counterparts in South Africa had told us that their country had the highest prevalence of FASD of any country in the world, but we were not convinced,” says Dr. Hoyme. However, it did not take many trips of meeting and assessing hundreds of children there for him and his fellow researchers to change their minds. “This is wine country with a climate similar to San Diego. Many families have been working in the wineries there for generations and it was not uncommon for them to even be partially paid with wine. As a result, many, many of them became alcoholic.”

These alcoholic mothers, who were likely to drink at all stages of their pregnancies, gave birth to unprecedented numbers of FAS children. Dr. Hoyme and his fellow researchers have found that



Dr. Hoyme has devoted much of his work to gathering data to help make FAS and FASD easier to diagnose. The four diagnostic categories he and his colleagues developed are now widely used in the U.S. and around the world.

about 8 percent of first graders in the Western Cape Province in South Africa, just outside of Capetown, have full-blown FAS.

“Our early research was focused on determining how common the condition was,” relates Dr. Hoyme. “More recently, we have looked at what can be done by way of intervention in the schools, what works and what doesn’t. Our most recent study is focused on prevention programs in order to reduce the rate of the condition. We are looking at the most effective ways to help mothers to stop drinking.”

MAKING THE DIAGNOSIS

During his 30 years of studying the effects of maternal alcohol use on fetal development, Dr. Hoyme has helped to more clearly define the spectrum of disorders that fall under the heading of FASD. Although the children first identified by Dr. Kenneth Jones as having FAS exhibited the tell-tale physical, behavioral and neurodevelopmental signs of the disease, continued research showed that many more children were affected by alcohol in utero, without showing all of the ‘signs’.

“It was discovered that there were children whose brain development was affected, but who lacked other features, such as a facial characteristics,” says Dr. Hoyme. “This came to be recognized as a question of timing. If the mother does not drink during the time that facial features are developing, but does drink earlier or later in her pregnancy, the child may end up with a normal face, but still

have developmental problems. These cases are harder to diagnose because you have fewer of the physical features to rely on.”

Recognizing that the first step toward possible life-changing intervention for FASD patients is accurate diagnosis, Dr. Hoyme and his research colleagues developed a set of diagnostic categories for use by primary care physicians. The categories and specific guidelines for using them in diagnosis were laid out in the journal *Pediatrics* in 2005 and now constitute a widely accepted revision of the Institute of Medicine’s official guidelines for diagnosis of FASD.

“Half of all children with FASD have an older sibling who also has the disorder.”

The four diagnostic categories named by Dr. Hoyme and his colleagues include FAS, Partial FAS, Alcohol-related Birth Defects (which can include things like heart and kidney problems), and Alcohol-related Neurodevelopmental Disorder, wherein a patient may demonstrate normal growth, normal head circumference and lack the typical facial findings, but have neurobehavioral problems characteristic of FASD. These categories are now widely used for FASD diagnosis not only in the U.S., but also in Africa, Europe, Eastern Europe, the Ukraine and Russia.

“The whole point of the research we are doing, and laying out

these categories, is so that a pediatrician can make a diagnosis of FASD as early as possible,” explains Dr. Hoyme. “You should not have to be a specialist to make this diagnosis. Our goal is to make this a condition that any physician can diagnose if he or she is careful and follows the guidelines.”

EARLY INTERVENTION IS KEY

Although FASD is not curable, as with many developmental disorders, early intervention is the key to a better life for patients and their families.

“In addition to their physical problems, these children tend to have learning problems, problems with judgment, ADHD, and problems with interpersonal relationships,” explains Dr. Hoyme. “If you know that FASD is the cause, there are things that can be done in the classroom and through early therapy that can help direct the course of that child’s life.”

Dr. Hoyme and his colleagues continue to research the most effective types of intervention for FASD, but other studies have already confirmed its value. Long-term studies in Scandinavia, where early intervention for FASD is mandatory, show that patients there did much better as adults than do many American FASD patients.

“The other reason to try to identify FASD and intervene as early as possible is that half of all children with FASD have an older sibling who also has the disorder,” says Dr. Hoyme. “That means that, if you could find those children and get to those mothers and help them with their drinking problem before they have another child, you could potentially prevent half of the cases of this disorder worldwide. That alone would be a tremendous impact.”

In addition to conducting global research, Dr. Hoyme remains an active clinician, seeing patients with FASD and other genetic disorders a half day a week in Sioux Falls and conducting clinics in Watertown, Pierre and Rapid City. He is currently the dysmorphologist on a large longitudinal study of infant mortality being conducted by Dr. Amy Elliott of Sanford and has recently received an Oxnard Foundation grant to study identification of FASD in early infancy.

It is difficult to quantify the extent of the problem of FASD in South Dakota. CDC estimates indicate that 57 percent of South Dakota women of childbearing age (18 to 44) drink alcohol and 19.4 percent admit to binge drinking, one of the highest levels in the country. Dr. Hoyme hopes to receive NIH funding to determine the prevalence of FASD in South Dakota,

which he suspects is higher than the national average. But despite the discouraging statistics, his message is one of hope to physicians, educators and parents.

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“If you know that FASD is the cause, there are things that can be done... that can help direct the course of that child’s life.”

they deserve a chance to live the best lives possible. If you give up on them, then you have lost a life. We hope that, through the work we are doing, their lives can be improved because, by doing so, you not only help that child and that family, but you also help society as a whole.” says Dr. Hoyme.

The CDC estimates the monetary lifetime cost of a single individual with FAS at \$2 million, putting the cost of FAS alone (not including other FASDs) at \$4 billion annually. Each day, from 6 to 22 infants are born with FAS in the U.S., and as many as 87 to 103 more are born with impairments related to prenatal alcohol exposure. The primary goal of Dr. Hoyme’s work is to eventually change those figures.

“I would love nothing more than, by the time I retire, to have had some impact on reducing the incidence of this condition around the world.”

South African students at one of the study sites for the Sanford-based Safe Passage study on infant mortality for which Dr. Hoyme is the dysmorphologist. The wine-growing regions of South African are plagued with FAS due to the high rate of alcoholism.



Sanford Consolidates in New Sanford Center



Sanford Health-MeritCare's research efforts, once spread out in multiple locations across Sioux Falls, are now located together inside the new Sanford Center. Sanford Health recently converted the former Hutchinson Technology building in northern Sioux Falls into corporate offices and space for all Sanford research centers. The Sanford Center was unveiled during a ribbon cutting celebration on June 8th.

"Consolidating our research efforts into one location allows

for greater collaboration and fast growth, hopefully leading to more discoveries and cures," says Dave Link, Senior Executive Vice President, Sanford Health-MeritCare.

The 300,000-square-foot Sanford Center is located near I-229 and I-90 in Sioux Falls. Currently, 64,000 square feet will be research administration and laboratory space including wet labs, offices and animal holding facilities (vivarium). Nearly 400 researchers and other employees will occupy the building, which will include all Sanford research centers:

- Cancer Biology Research Center
- Cardiovascular Health Research Center
- Health Disparities Research Center
- Sanford Children's Health Research Center
- Methodology and Data Analysis Research Center
- Women's Health Research Center
- National Institute for Athletic Health & Performance at Sanford
- The Sanford Project – Curing Type 1 Diabetes Through Beta Cell Regeneration

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"Our researchers continue to be published and recognized for their discoveries, and Sanford Research/USD offers exciting challenges both in well-established research projects and ever expanding research opportunities," said Ben Perryman, PhD, President, Sanford Research/USD. "As a result, we anticipate accelerated researcher and physician recruitment for our entire health system. This facility puts Sanford research recruitment capacity some three to five years ahead of schedule."

The Sanford Center will also hold the new Sanford Health-MeritCare Sioux Falls corporate headquarters for leaders from both Sioux Falls and Fargo as well as other corporate functions including Executive Administration, Sanford World Clinic, Health Policy, Public Affairs and Research Park Development, among others.