Biomedicine

Lisa Seachrist reports from Minneapolis at a meeting of the American Society of Human Genetics

Birth defects too often blamed on alcohol

Physicians and geneticists too readily blame birth defects on a mother's use of alcohol during pregnancy and may miss other genetic causes of the abnormalities, says an Arizona researcher. In a study of children previously identified as suffering from the effects of fetal exposure to alcohol, he found that 13 percent suffered from misdiagnosed genetic problems.

"Some of these children were labeled as suffering from mild cases of fetal alcohol effects, when in fact they have something entirely different," says H. Eugene Hoyme of the University of Arizona Health Sciences Center in Tucson. He urges doctors and geneticists to eliminate the diagnosis of fetal alcohol effects (FAE), considered a mild form of fetal alcohol syndrome.

First described in 1973, fetal alcohol syndrome, one of the most common causes of birth defects, occurs in 1 in 500 to 1 in 1,000 births. Children with the syndrome typically have smaller heads, small eye openings, flattened noses, and smooth upper lips. They also tend to be short and to have low IQs.

Diagnosis of the disorder hinges on facial abnormalities, short stature, and low IQ, says Hoyme. But children who don't meet all three criteria are often diagnosed with FAE, on the assumption that their birth defects arose from their mothers' drinking during pregnancy.

Some scientists, however, consider FAE such a catchall diagnosis that it obscures some important genetic diseases. Hoyme and his colleagues studied 437 Arizona children, 19 percent of whom had been diagnosed with fetal alcohol syndrome. The rest had diagnoses ranging from FAE to probably FAE to some FAE.

The team reanalyzed the cases, this time limiting the definition of fetal alcohol syndrome to the characteristic facial abnormalities. With this revision, the researchers diagnosed fetal alcohol syndrome in a whopping 56 percent of the children.

But they also found that 13 percent suffered from conditions such as Down's syndrome and neurofibromatosis—conditions doctors had previously failed to diagnose. The researchers classified 41 percent of the children as having had some prenatal alcohol exposure, but they could not link the birth defects to alcohol. Some of the children had two diagnoses.

"Any abnormality is now described as an effect of alcohol if the mother drank at all during pregnancy," says Hoyme. Even though drinking during pregnancy is a problem, "we risk stigmatizing children and missing other diagnoses which require medical follow-up if we too easily point to alcohol."

Racial differences in tumor disorder

Two people with an identical genetic defect may suffer widely different medical problems. In the case of neurofibromatosis, researchers from Washington, D.C., and Ohio have found that whites are more likely than blacks to suffer from benign, yet potentially dangerous, optic nerve tumors.

"What this tells us is that single-gene disorders manifest themselves with respect to the other genes in individual carriers," says Howard M. Saal of the Children's Hospital Medical Center in Cincinnati.

Saal and colleagues at the Children's National Medical Center in Washington, D.C., studied 372 children with neurofibromatosis, a disease marked by tumors of the skin and central nervous system and skin pigmentation known as café au lait spots. Using magnetic resonance imaging (MRI), the team found optic nerve tumors, which can cause blindness and pituitary problems, in roughly 1 out of 5 white children but in only 1 out of 62 black children.

Saal says the finding illustrates how differences in other genes can affect neurofibromatosis. However, he recommends MRI scans for all children with the disease, because 7 to 10 percent suffer from malignant brain tumors regardless of race.

Paleontology

Richard Monastersky reports from Pittsburgh at the annual meeting of the Society of Vertebrate Paleontology

Squeezing blood from a stone

By digging into the marrow of a *Tyrannosaurus rex*'s bones, scientists have discovered what appear to be organic molecules from the dinosaur's blood. If these compounds survived 65 million years of entombment against all odds, they might answer major questions about the evolution of these ruling reptiles.

Mary H. Schweitzer of Montana State University in Bozeman and her colleagues collected their samples from well-preserved dinosaur tissue. The marrow seems to have escaped the typical fossilization process, in which minerals replace organic molecules.

To analyze compounds from the marrow, the scientists used six independent techniques, including nuclear magnetic resonance and high-performance liquid chromatography. They also injected extracts from the marrow into rats to see whether they produced antibodies in response to the foreign material.

All lines of evidence suggest that the *T. rex* compounds are proteins containing a heme group—a ring-shaped molecule surrounding a central iron atom. Because red blood cells have heme in the form of hemoglobin, the findings raise the possibility that fragments of blood protein have survived in the *T. rex* marrow. If so, scientists could use the amino acid sequence in the proteins to determine whether dinosaurs are more closely related to birds or to crocodiles.

But first, Schweitzer and her colleagues must overcome the skepticism of molecular biologists, who believe that organic material cannot survive for millions of years.

James Garey, a molecular biologist at Duquesne University in Pittsburgh, says he is convinced that Schweitzer has found heme. But he wonders about its origin.

Although hemoglobin only exists in higher animals, almost every living creature has heme in its cells, says Garey. "Maybe some bacteria grew in there later. Maybe it's heme from their [cells]," he suggests.

Proof would come from the proteins. If Schweitzer and her coworkers can find and sequence the amino acids, they could rule out possible contamination from bacteria, says Garey.

A most dangerous sawfish

Modern sawfish don't fit their name. Their long snouts, covered with a few dozen widely separated teeth, look more like combs than anything else.

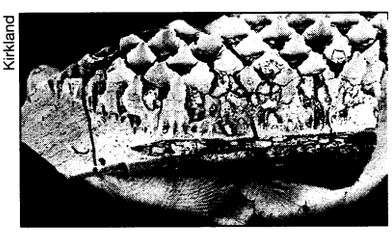
But ancient sawfish practiced truth in advertising. Two paleontologists report finding hundreds of sharp, tiny teeth lined up along the nose of a Cretaceous sawfish called *Schizorhiza*.

"This creature went to the extreme. It had a perfect saw," says James I. Kirkland of Dinamation International Society in Fruita, Colo.

Although paleontologists first described the genus *Schizorhiza* in 1930, they did not know its tooth arrangement until last year. At that time, Martha C. Aguillon Martinez of the Secretary of Public Education of Coahuila discovered a *Schizorhiza* snout in northern Mexico.

Kirkland and Aguillon Martinez report that *Schizorhiza* had more than 200 closely spaced teeth along its nose. An additional 1,600 or so teeth grew under the skin, ready to replace the ones on the surface. While modern sawfish sometimes use their snouts for probing seafloor sediments, *Schizorhiza*'s saw was a specialized tool for hunting and would not have worked for sifting the ocean bottom.

Kirkland



Fossil impression of Schizorhiza *teeth.*