spotlight on health

Juvenile Paget's Disease: Rare Childhood Bone Disorder

(NAPSA)—Two seemingly unrelated Navajo children have one painful thing in common: juvenile Paget's disease (JPD), an extremely rare bone metabolism disorder. Now, researchers at the Shriners Hospital for Children and Washington University School of Medicine, both in St. Louis, have discovered that the two patients also share an unusual genetic defect.

The research team found that both patients are completely missing the gene for a recently discovered protein called osteoprotegerin (OPG) known to protect bone. The study is the first to identify a genetic cause for JPD and was published in the July 18, 2002, issue of the *New England Journal of Medicine.*

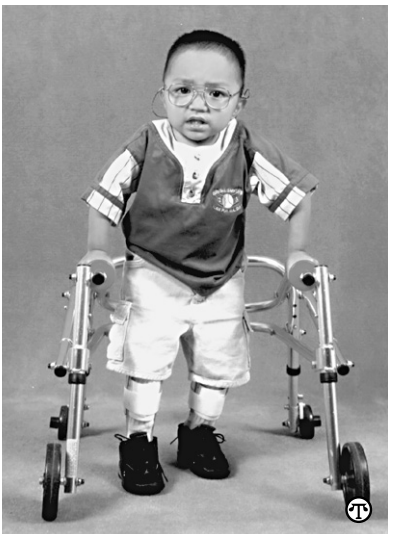
"By identifying this genetic defect in two people, our results not only provide insight into the cause of JPD, but also shed light on the control of bone metabolism in general," said lead investigator Michael P. Whyte, M.D., director of the Center for Metabolic Bone Disease and Molecular Research at the St. Louis Shriners Hospital.

JPD has only been reported in about 40 people worldwide. It is a painful skeletal disease characterized by abnormally fast formation and breakdown of bone throughout the body, resulting in debilitating fractures and deformities beginning soon after birth.

Because JPD can occur among siblings, it is presumed to have a genetic basis. Until now, that genetic defect was unknown.

The research team examined DNA samples from two Navajo patients. The first was referred to the St. Louis Shriners Hospital from New Mexico in 1996 for confirmation of diagnosis and treatment at one year of age. The team later learned that a second JPD patient, described in the medical literature in 1979, also was living in New Mexico.

Thanks to simultaneous advances in the Human Genome Project, centered in part at Washington University, the team was able to pinpoint exactly where DNA had broken off in these two patients.



Shriners Hospital patient Anfernee is one of only 40 people who has juvenile Paget's disease.

The St. Louis discoveries illustrate the critical importance of OPG in regulating bone formation and breakdown (turnover) in humans. It is now possible to detect carriers of the disease and diagnose it prenatally in the Navajo population. Importantly, the observations suggest that an injection of OPG, perhaps every few weeks, might be an effective therapy for JPD in Navajo children.

For more information on Shriners network of 22 hospitals that provide medical care and services totally free of charge to children with orthopaedic problems, burns and spinal cord injuries, write to: Shriners International Headquarters, Public Relations Dept., 2900 Rocky Point Dr., Tampa, FL 33607, or visit the Web site at www.shrinershq.org. Treatment is provided to children under age 18 regardless to race, religion or relationship to a Shriner.

If you know a child Shriners can help, call 1-800-237-5055 in the United States or 1-800-361-7256 in Canada.