Gene Therapy for Morquio A Syndrome Show Promise in Slowing Progression of Disease

Findings Announced at Nemours-sponsored conference in August

Wilmington, DE – A team led by Shunji Tomatsu, MD, PhD, principal research scientist with Nemours Biomedical Research, has been conducting early stage investigation on a gene therapy for Morquio A syndrome. Preliminary data in mice models suggest that this approach could slow or prevent the progression of disease in bone and cartilage.

> *“Nemours/Alfred I. duPont Hospital for Children is renowned for its clinical and research expertise in Morquio syndrome and has been caring for Morquio patients for 30 years. The hospital actively treats more than 70 patients with Morquio, many of whom attended the recent conference hosted by Nemours.”*

About Morquio syndrome

Morquio syndrome Type A is a rare lysosomal storage disease characterized by short stature, a large head, severe scoliosis, cervical spinal cord compression, tracheal obstruction, distinctive facial features and in some cases, heart, liver and vision problems.

Morquio A syndrome is estimated to occur in one in 250,000 births worldwide. About 500 people in the U.S. are known to have Morquio A syndrome. The disease may not be apparent at birth; symptoms may become noticeable between ages 1 and 3 and get worse as the child grows.

Morquio A syndrome is part of a group of diseases called mucopolysaccharidoses (MPS). In children with Morquio A syndrome, the body cannot break down sugar chains called glycosaminoglycans (GAGs) that are present in bone, cartilage, eye corneas, skin and connective tissue (tendons, ligaments, etc.) As a result, GAGs collect in cells, blood, and connective tissue and cause damage over time. In Morquio A syndrome, the enzyme called galactosamine-6-sulfatase is missing. The damage caused by the lack of the enzyme affects a child’s appearance, tissues, bones, organ function and physical abilities. Bone problems can lead to major health problems. For example, the small bones at the top of the neck may slip and injure the spinal cord, causing paralysis. Surgery to correct problems such as these should be done if possible.

In addition to surgery, the gold standard of treatment for Morquio A syndrome has been enzyme replacement therapy (ERT). However, ERT is expensive, involves a weekly five-hour intravenous infusion, and has limited impact on patients’ severe bone pathology.

Promising results

Dr. Tomatsu’s research showed that enzyme activity was elevated in treated mice, resulting in complete clearance of GAG storage in certain cells and reduced storage in others. The high enzyme levels combined with longer circulation time could have led to increased penetration into bone and cartilage thereby improving storage in those regions. The results of these data have been presented at the Fourth Morquio Conference (July 29; Nemours) and at the 15th Annual International Symposium on MPS and Related Diseases (August 2-4; San Diego, CA).

Nemours/Alfred I. duPont Hospital for Children is renowned for its clinical and research expertise in Morquio syndrome and has been caring for Morquio patients for 30 years. The hospital actively treats more than 70 patients with Morquio, (including Carly, inset) many of whom attended the recent conference hosted by Nemours.

About Nemours

Nemours is an internationally recognized children's health system that owns and operates two free-standing children’s hospitals: the Nemours/Alfred I. duPont Hospital for Children in Wilmington, Del., and Nemours Children's Hospital in Orlando, Fla., along with outpatient facilities in five states, delivering pediatric primary, specialty and urgent care. Nemours also powers the world’s most-visited website for information on the health of children and teens, KidsHealth.org, and offers on-demand, online video patient visits through Nemours CareConnect. Nemours ReadingBrightstart.org is a program dedicated to preventing reading failure in young children, grounded in Nemours’ understanding that child health and learning are inextricably linked, and that reading level is a strong predictor of adult health.

Established as The Nemours Foundation through the legacy and philanthropy of Alfred I. duPont, Nemours provides pediatric clinical care, research, education, advocacy and prevention programs to families in the communities it serves.

For further information: Karen Bengston 302-293-4928 karen.bengston@nemours.org

---

Additional assets available online: **Photos** (1)

https://nemours.mediaroom.com/news-releases?item=122708