“Our goal is to make our patients as functional and independent as possible.”

William Mackenzie, MD  
Chairman – Orthopedics

For information, please call Angela Duker at (302) 651-4181 to learn more about our program.

To schedule an appointment, please call (302) 651-5916

The Nemours/Alfred I. duPont Hospital for Children offers a treatment philosophy and level of care and comfort that sets the standard for children’s health care. Nestled among beautifully landscaped gardens, lush lawns, sunlight, and fresh air, the 200-bed teaching hospital offers pediatric specialty care in a patient- and family-centered environment.

What is Skeletal Dysplasia?

In the human body, there are 206 bones that form the skeleton. When the bones and the cartilage in the skeleton do not form or develop normally, they are said to be dysplastic. The skeletal dysplasias are a group of medical conditions in which the bones of the skeleton are abnormal.

Dysplasias are characterized by abnormal growth, development, shape, or integrity of the bones. They can lead to a wide variety of medical problems, but the most common feature among them is dwarfism (defined as having a final adult height below 4’10”).

Children with skeletal dysplasias have multiple medical and social needs, many of which are unique to their condition.

The Nemours Difference

The Skeletal Dysplasia Program at the Nemours/Alfred I. duPont Hospital for Children is one of the world’s foremost centers in the treatment of skeletal dysplasia, offering expert, state-of-the-art medical and surgical care for children with dwarfism.

Together we have more than 75 years of experience in the diagnosis and management of children with skeletal dysplasia.

It is comforting for parents to know that their child’s complex medical and surgical needs can be met in one place. If your child needs to see multiple specialists, we share our findings with each other so that treatment is a true collaboration.

Evaluation and Treatment

The initial visit to the skeletal dysplasia clinic generally includes a diagnostic visit performed by Michael Bober, MD, PhD. These visits are quite comprehensive and can often take one to two hours to complete. During this appointment he will:

- review medical and family history
- examine previous X-rays and scans
- perform physical examination including detailed measurements
- assess ranges of joint motions and relative length of limb segments
- determine if further radiological, biochemical, or molecular testing is necessary

A specific diagnosis of a skeletal dysplasia in an individual is based upon the totality of all the data obtained. Determining a diagnosis can often be accomplished in this initial evaluation, but sometimes this is not the case. Some dysplasias can take time to diagnose, and this type of comprehensive evaluation may need to be repeated. These disorders can evolve and change with time.

Integrated System of Care

Once a specific diagnosis is made, the treatment and management strategies are tailored to meet the needs of the patient and their family. In addition to care and referrals, emotional and psychosocial support and genetic counseling services are provided. Orthopedic care is a critical component in the effective management of patients with skeletal dysplasias. Treatment is directed toward maximizing mobility, correcting deformity, and preventing complications.

In the genetics clinic, Dr. Bober follows patients at approximately six-month intervals until the age of six. Then visits are typically spaced yearly. William Mackenzie, MD, will make follow-up determinations based upon their most recent evaluation. Our program is structured so that a family can come and see these physicians and any other specialists in the same visit.

For families traveling long distances, the Ronald McDonald House of Delaware is located adjacent to the Nemours/Alfred I. duPont Hospital for Children. Visit www.rmhde.org or call (302) 656-HUGS.

Meet Our Team

William Mackenzie, MD
Chairman – Orthopedics
Chair – Little People of America Medical Advisory Board
Medical Director – Dwarf Athletic Association of America

Michael Bober, MD, PhD
Geneticist
Co-Director – Skeletal Dysplasia Program
Member – Little People of America Medical Advisory Board
Member – Potentials Foundation Medical Advisory Board
Member – Walking with Giants Medical Advisory Board

Charles Scott, MD
Senior Geneticist and Consultant
Co-Director – Skeletal Dysplasia Program
Member – Little People of America Medical Advisory Board

Colleen P. Ditro, CPNP, MSN, RN
Pediatric Nurse Practitioner
President – Pediatric Orthopaedic Practitioner Society
Member – National Association of Pediatric Nurse Practitioners

Angela L. Duker, MS, CGC
Genetic Counselor
Member – Potentials Foundation Medical Advisory Board
Member – Walking with Giants Medical Advisory Board