

Highlights from the Baltimore Clinical Care Conference

The 5th Clinical Care Meeting on osteogenesis imperfecta was held November 6-8, 2013 in Baltimore, MD. Chaired by Dr. Jay Shapiro from the Kennedy Krieger Institute and sponsored by the OI Foundation and the Charitable Research Foundation, the meeting brought together a group of physicians who are experienced in caring for people who have OI. The meeting had three major goals – to identify useful information that is emerging from basic and clinical research, to make that information easily available to medical practitioners and to identify areas in clinical care that need to be studied.

Abstracts that summarize each speaker's presentation have been published in the March 2014 on-line medical journal, **Journal of Musculoskeletal and Neuronal Interactions**.

The presentations covered a wide variety of clinical topics connected to the diagnosis, treatment and management of OI in children and adults. The large number of topics reflects the growing awareness that OI is a complex disorder that affects many parts of the body besides the skeleton. A summary of main ideas follows.

Gene Therapy – Stem cell therapy is a potential treatment for OI. While progress has been made in the technology in the lab, data is lacking on how to effectively deliver the new cells into the person who has OI.

Dental Care – The effect of OI on teeth goes beyond dentinogenesis imperfecta and includes problems with growth of the face, head, and jaw. People with moderate and severe OI tend to have more serious issues in this area. In some cases both orthodontia and orthognathic surgery may be necessary.

Pregnancy – Many women who have OI and short stature have experienced successful pregnancies, but the normal physiologic changes of pregnancy can lead to pulmonary and skeletal problems for some women who have OI. Pregnancy can pose a high degree of risk for women with any cardiac dysfunction.

Hearing Loss – OI related changes to type 1 collagen lead to changes in the structures of the inner ear. Hearing tests should begin during school years. Bone anchored hearing aides are not beneficial but cochlear implants with modifications are a viable option.

Eye Related Care – People with OI frequently experience common vision problems such as nearsightedness. OI also increases the fragility of the tissues of the eye, may increase the odds of developing glaucoma and retinal tears and makes treatment for these complicated problems more difficult.

Gastrointestinal Issues – For many people, OI is accompanied by gastrointestinal symptoms that include constipation and abdominal pain. In the absence of clear data, care is primarily supportive. Research is needed into causes, role of OI type, and response to treatments in children and adults.

The Skin – Alterations in the quantity and quality of type 1 collagen causes skin thinness, a tendency to form scars and other problems. Current treatment focuses on gentle skin care, and sun protection.

Pulmonary Concerns – Two elements (abnormal chest wall shape and altered lung tissue) combine to increase the risk for serious breathing problems for people who have OI. Short stature, scoliosis, and long bone deformities increase the difficulty of diagnosis and treatment of breathing problems.

Heart Disease – Cardiovascular diseases among people with OI is increasingly recognized as life-threatening and treatable. The exact prevalence is unknown, but valve diseases, aortic disease and arterial dissection have been identified, as well as related conditions such as hypertension (high blood pressure). When surgery is needed, seeking treatment at specialized centers with experience in surgery and physical rehabilitation for people with connective tissue disorders will reduce the potential for complications.

Basilar Invagination – Basilar impression, cranial settling and basilar invagination are three different problems that are seen in people who have OI. Deformity of the cervical spine and compression of the brainstem from these conditions can lead to symptoms that may become life threatening. At this time, surgical intervention is difficult.

Orthopedic Problems and Treatments – A series of presentations were given on specific orthopedic issues. There have been steady advances in the treatment of long bone deformities in children with OI. The use of medical therapies (drugs) improves the bone but does not provide normal bone density and function. Indications for surgery include frequent fractures (despite optimum medical treatment) and severe bowing. The age of the child, size of the bone and degree of bowing are factors in the selection of IM rod. Surgery to correct bowing of the humerus often improves comfort and the child's ability to use the arm. Spinal deformities are frequent in OI and the prevalence of scoliosis is much higher in OI than in the general population. Scoliosis in children with Type III and IV OI gets worse much quicker than in children with Type I OI. Joint replacement especially of the knee and hip are increasingly considered for adults who have OI. Poor bone quality, small size, the presence of rods in adjacent long bones, and fracture history are factors that complicate these surgeries. Ligament laxity has serious implications for the long-term stability of the joint replacement. Complications related to all orthopedic surgeries are common and should be discussed with the surgeon prior to deciding on a treatment.

Rehabilitation and Physical Activity – Rehabilitation plays a crucial role in promoting health and well-being for the person who has OI. At every stage of life, carefully designed programs for physical activity can help prevent obesity, improve mobility, aerobic fitness and independence as well as reduce pain. Weight-gain in teens and adults is a significant cause of mobility loss.

Bisphosphonate Treatment – A series of talks were given on this topic. Treatment with intravenous bisphosphonate has had a beneficial effect in many OI children; decreasing fractures and pain, and increasing mobility. A comparison of two bisphosphonates – pamidronate and zoledronic acid—found similar effects on fracture incidence. Oral bisphosphonate drugs do not appear to be consistently effective in children. Not all children continue to respond to bisphosphonate therapy as they enter adulthood.

This article appeared in the Summer 2014 issue of the OI Foundation newsletter, ***Breakthrough***. It was written by Mary Beth Huber, OI Foundation Director of Program Services, and reviewed by Dr. Jay Shapiro, Kennedy Krieger Institute, Baltimore, MD.