

Where does pain come from in XLH hips and knees?

A joint like the hip or knee are structures that are dependent on many tissues designed for stability and the integrity of function.

These joints are intended for weight-bearing and motion, and depend on elements of the joint structure to distribute weight (cartilage, synovial fluid), bone (shape), muscles (stabilize, permit motion) and ligaments (stabilize).

In metabolic bone disease, there are several issues that may lead to later complications in the adult,

1. abnormal mineralization with softening/bowing
2. abnormal tethering of muscles (tendons) and ligaments to bone
3. abnormal shaping of the bones

In the adult skeleton, pain may come from these, and may be complicated by inflammation, by overgrowth of bone causing impingement on motion and early osteoarthritis. When the joint fails entirely, we consider a joint replacement. Before this surgical intervention, we often approach these problems like every other case of osteoarthritis. Our goal is to define how to approach XLH joints and musculoskeletal complications better.

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Why do I have back pain?

Back pain occurs in 90% of the population at some time in our lives. Sometimes it is from a herniated disc, but most of the time there is not a single, clear anatomical reason for back pain. The discs wear out, the back stiffens, facet joints which permit vertebral body motion get arthritic, ligaments thicken... we get shorter/stiffer over time. Sometimes there may be further mechanical problems in the skeleton that increase the work through the spine. In XLH adults, all of these things may occur affecting function and comfort, and they may be worsened by ligament overgrowth with calcium or bone. Stretched muscles (back strain) and ligaments (sprain) may play a role, too. And there is some understanding that chronic pain has its own life and patterns in individuals with longstanding disease. It is not clear whether treatment for the XLH will improve or worsen some of these musculoskeletal problems.

My interest is to discover those aspects of XLH that impair the functional status and cause pain in the adult patient. I approach this as a rheumatologist, depending on the insights of colleagues in genetics, pediatrics and endocrinology. –Dr. M. Seton, panelist XLH Day