Children with Down syndrome at increased risk for inflammatory, erosive arthritis

10 November 2019

A new study finds that children with Down syndrome are at an increased risk of an associated form of arthritis. Additionally, researchers recommend changing the name to Down syndrome-associated arthritis to more accurately reflect the inflammatory and erosive nature of the condition. Details of this study will be presented at the 2019 ACR/ARP Annual Meeting (Abstract #1817).

About one child in every 1,000 develops some type of chronic arthritis. These disorders can affect children at any age, although rarely in the first six months of life. It is estimated that around 300,000 children in the United States have been diagnosed with the condition. Down syndrome is a condition in which a person has an extra chromosome number 21, which affects how the body and brain develop.

Arthropathy of Down syndrome has an increased incidence and prevalence compared to Juvenile Idiopathic Arthritis (JIA). However, the disease is rarely recognized at onset and remains under-diagnosed. Children with arthropathy of Down syndrome often present with significant joint damage and disability at diagnosis. A group of researchers from Ireland conducted a cross-sectional, observational study to identify undiagnosed cases of arthropathy of Down syndrome, document the time to diagnosis among these patients, and describe the clinical, laboratory and radiological features of the condition at diagnosis.

"Given the paucity of information in the literature with regards to arthritis in children with Down syndrome, our initial aims were to identify whether arthritis in Down syndrome is missed leading to a delay in diagnosis, describe the clinical and radiological features of inflammatory arthritis in children with Down syndrome, and estimate the prevalence of inflammatory arthritis in children with Down syndrome," said Charlene M. Foley, MBBS, BSc, Ph.D., a clinical researcher at the National Centre for Pediatric Rheumatology at Our Lady's Children's Hospital in Crumlin, Ireland, and the study's lead author. "If we know there is increased risk in children with Down syndrome, we are more likely to consider it as a possible diagnosis. Early recognition leads to earlier instigation of appropriate treatment and, therefore, better clinical outcomes and quality of life for a population of children already at risk of a number of co-morbidities that can impact their lives."

Researchers invited children (zero to 21 years old) with Down syndrome to participate in a musculoskeletal screening clinic where they received a detailed examination from a pediatric rheumatologist. A subsequent clinical visit with a different physician confirmed all suspected cases of arthropathy of Down syndrome. Physicians instigated investigations and treatment following normal clinical practice for JIA. The researchers collected data on a convenience sample of 21 newly diagnosed children with JIA to create a comparison group.

Over an 18-month period, 503 children with Down syndrome were screened for arthritis, with 18 new cases diagnosed. In total, the study identified 33 children with arthropathy of Down syndrome, combining cases that predated the study's commencement and those children referred to the center during the study period. The study's results suggest that prevalence of arthropathy Down syndrome is 20 per 1,000 children with Down syndrome.

The researchers also observed significant delays in diagnosis of arthritis in children with Down syndrome. The majority of children presented with polyarticular, RF-negative arthritis, with a predominance of arthritis in the small joints of the hands and wrists. No children with arthropathy of Down syndrome in the study were ANA positive. Erosive changes were reported on X-ray in more children with arthropathy Down syndrome (42 percent) than the JIA group (14 percent). Future

research in this patient population may help accurately define disease pathogenesis, identify disease biomarkers and establish best practices for treatment, the researchers concluded. They also suggest that "Down syndrome-associated arthritis" would be a more accurate term than "arthropathy of Down syndrome."

"To our knowledge, this is the first study to consider screening children with Down syndrome for arthritis. Through this simple, non-invasive process we detected a number of undiagnosed cases of Down syndrome associated arthritis (DA)" said Dr. Foley. "We observed a high degree of methotrexate-associated side effects in children with DA. With this knowledge, clinicians may consider altering their treatment choices in favor of biological therapy for this cohort of children. Our study highlighted that the clinical phenotype of the condition is inflammatory and erosive in nature. Our proposal to rename the condition Down syndrome-associated arthritis is to reflect the inflammatory, erosive nature of the disease."

More information: Study: Arthropathy of down Syndrome: An Under-diagnosed Inflammatory Joint Disease That Warrants a Name Change

Provided by American College of Rheumatology