Down syndrome arthropathy diagnosis delayed a year, optimal treatments still unclear

November 10 2019

A new study found that patients with Down syndrome arthropathy continue to have an approximate year-long delay in diagnosis from the onset of their symptoms, and that optimal therapy for this condition remains unclear (Abstract # 2722). Details of this research will be presented at the 2019 ACR/ARP Annual Meeting in Atlanta.

Down syndrome arthropathy is under-recognized, with an average 19-month delay in diagnosis. The majority of patients present with polyarticular, rheumatoid factor and antinuclear antibody (ANA) negative disease. Current juvenile idiopathic arthritis (JIA) therapies appear to be poorly tolerated, more toxic and less effective in patients with the condition. This observational study's objective was to characterize the clinical manifestations and therapeutic preferences in Down syndrome arthropathy using the Childhood Arthritis and Rheumatology Research Alliance (CARRA) registry. CARRA's large multicenter observational registry began collecting data on children with JIA in the United States and Canada in July 2015. Down syndrome is documented in the CARRA registry as a coexisting condition. "Down syndrome is one of the most common birth conditions in the United States and has a variety of associated complex medical challenges, such as increased incidence of oncologic and autoimmune conditions," says Jordan T. Jones, DO, MS, a pediatric rheumatologist at Children's Mercy Kansas City and assistant professor of pediatrics at the University of Missouri-Kansas City School of Medicine, and the study's lead author.

"Additionally, children with Down syndrome are at risk for delayed motor skills and functional limitation. Due to this, their symptoms can be overlooked or attributed to something other than arthritis. We hope to raise awareness and improve screening so there is a decrease in the delay in diagnosis. After diagnosis, we have to determine optimal therapy as these patients do experience more medication toxicity, intolerance and ineffectiveness compared to those with JIA." The researchers identified 36 patients with Down syndrome arthropathy in the total population of 7,337 JIA patients in the registry. Musculoskeletal symptoms began around seven years of age and there was an average delay of 11.5 months to arthritis diagnosis. At diagnosis, 64 percent of the patients had a polyarticular, RF-negative presentation, 39 percent reported morning stiffness with an average of four active and four limited joints, and 27 percent had elevated inflammatory markers.

Over half (64 percent) the patients began a disease-modifying antirheumatic drug (DMARD) at diagnosis, with 36 percent simultaneously starting a biologic therapy. Over the course of the disease, 78 percent used a DMARD, with all using methotrexate at some point, and 75 percent used a biologic, mostly etanercept. Of those patients exposed to DMARD therapy, more than half (54 percent) had at least one DMARD change, mostly due to methotrexate adverse effects. However, at the last recorded visit, 42 percent of patients were on methotrexate. Fifty-five percent of patients had at least one change in biologic therapy due to inadequate response. Between the first and last visit, significant improvements in active joint count and physical global disease activity were seen, but there was still a substantial disease burden noted.

The study's findings show that patients with Down syndrome arthropathy continue to experience an approximate year-long delay in diagnosis after symptom onset. Additionally, optimal therapy for Down syndrome arthropathy remains unclear, and current barriers include DMARD

intolerance and anti-TNF effectiveness, the study shows.

"There is still a delay in recognition of Down syndrome arthropathy, which means we need more education and awareness for this disease, focused on patients, families and providers that see children with Down syndrome prior to a rheumatologist," says Dr. Jones. "We have proposed regular screening and earlier referral if there are concerns for arthritis in a child with Down syndrome. Additionally, once a child is diagnosed with Down syndrome arthropathy, we need to determine optimal therapy with the least amount of adverse effects and toxicity to improve outcomes."

More information: Study: The down Syndrome Arthropathy Cohort in the New Childhood Arthritis and Rheumatology Research Alliance (CARRA) Registry: Clinical Characteristics, Treatment and Outcomes Pediatric Rheumatology - Clinical

Provided by American College of Rheumatology