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MSUS ACADEMY

NEWSLETTER

CPPD in Down syndrome

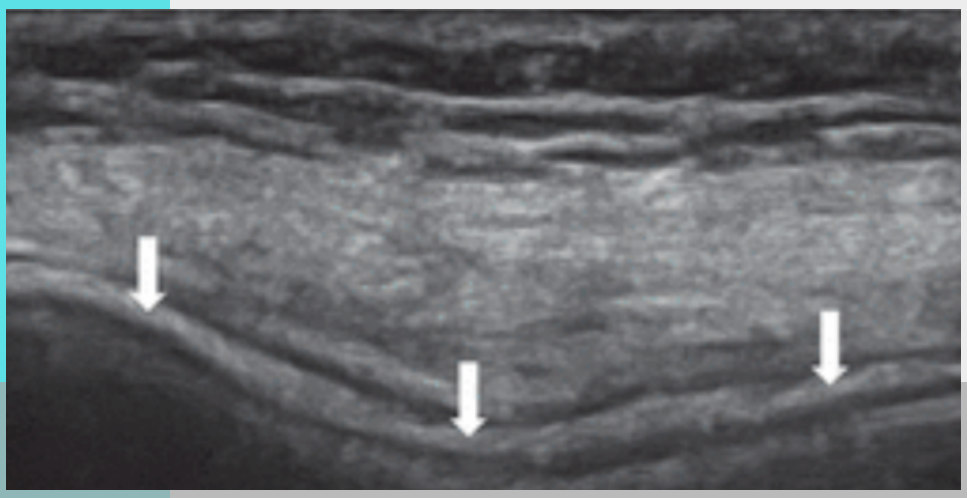
presented by
Dr. Christian Marx

We present a rare form of calcium pyrophosphate dihydrate disease in Down syndrome.
In the current case, structural and typical lesions were found in the form of chondrocalcinosis of fibrous and hyaline cartilage (figure below, hyaline cartilage of the femoral condyles).
The findings have a high sensitivity (80%) and specificity (>93%).
In the young patient, chondrocalcinosis was diagnosed by sonography, and following positive synovial analysis with crystal detection, a diagnosis of calcium pyrophosphate deposition disease (CPPD) was made.

Further readings (free article):

Article link

CPPD - The Lancet Rheumatology



Interview with
Dr. Christian Marx

Dr. Christian Marx
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Rheumatology and owner at RZO - Rheumatology in Zurich Oberland, Uster, Zurich, Switzerland

MSUS Academy: You have been president of the musculoskeletal section of the SGUM in Switzerland for many years. What is your role, and what are the challenges within the organization and externally?

CM: As president of the musculoskeletal section of the Swiss Society for Ultrasound in Medicine (SGUM), and Rheumatologist my role is primarily to motivate, coordinate, and inspire — fostering enthusiasm for musculoskeletal ultrasound as a powerful and dynamic diagnostic tool. A central challenge lies in engaging motivated colleagues who contribute voluntarily, without financial or career incentives, while ensuring the inclusion of all Swiss language regions to create a truly national and collaborative platform.
My focus is on maintaining and advancing quality and educational standards: continuously updating learning objectives to reflect new scientific and technical developments, training highly skilled tutors and supervisors, ensuring fair and competency-based examinations, and expanding the course system with consistent quality control. I also promote the creation of educational resources such as learning videos and concise manuals to support trainees effectively.
Beyond education, I aim to strengthen the organizational and administrative infrastructure, improve online visibility, and provide a professional home for all physicians engaged in musculoskeletal ultrasound. Externally, a key challenge is to secure fair reimbursement structures that reflect the value of high-quality training and practice within the Swiss tariff system.
Representation within the national umbrella organization is another vital part of my role — ensuring that the achievements of the musculoskeletal section are recognized and supported at a higher level.
Finally, I work to build international collaborations with like-minded experts while continuing to invest in my own professional growth and developments.

https://www.msus-schweiz.ch/

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MSUS Academy: You published a case of severe CPPD in a young patient with Down syndrome [https://econtent.hogrefe.com/doi/epdf/10.1024/1661-8157/a002334]. When we diagnose CPPD in adolescents, what causes should we consider?

CM: CPPD in adolescence is an absolute rarity and should always raise suspicion of an underlying genetic or metabolic disorder. While the disease typically manifests after the age of 50, cases in adolescents or young adults are almost always secondary.
The most important causes include hereditary forms, particularly mutations in the ANKH gene, which are inherited in an autosomal-dominant manner and lead to increased extracellular pyrophosphate transport, thereby promoting crystal formation.
In addition, metabolic and endocrine disorders play a central role: hemochromatosis, hyperparathyroidism, hypomagnesemia (e.g., in Gitelman or Bartter syndromes), hypophosphatasia, chronic kidney disease, and hypothyroidism are the main differential diagnoses. These conditions alter pyrophosphate metabolism or cartilage homeostasis, thus facilitating calcium pyrophosphate deposition. Syndromic causes have also been described, particularly in Down syndrome, where altered matrix and cartilage biology appear to contribute. Individual case reports have also described associations with Wilson's disease and ochronosis.
The basic diagnostic work-up for CPPD in patients under 50 years of age should therefore always include laboratory tests for calcium, phosphate, magnesium, alkaline phosphatase, ferritin, creatinine, TSH, and parathyroid hormone, supplemented by imaging (ultrasound or radiography) and, if indicated, synovial fluid analysis to confirm crystal presence. If no metabolic cause is identified, genetic testing for ANKH mutations is warranted.
In summary, CPPD in young individuals is almost never idiopathic. Potential causes include familial forms due to ANKH mutations, metabolic or endocrine disorders (especially hemochromatosis, hyperparathyroidism, hypomagnesemia, hypophosphatasia, hypothyroidism, and renal insufficiency), and syndromic conditions such as Down syndrome. The diagnosis of "idiopathic CPPD" in this age group should only be made after exclusion of these underlying causes.

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MSUS Academy: What is the best technique for examining the lateral hip in GTPS? [https://econtent.hogrefe.com/doi/10.1024/1661-8157/a002241?url_ver=Z39.88-2003&rfr_id=ori:rid:crossref.org&rfr_dat=cr_pub%20%200pubmed]?

CM: The optimal ultrasound technique for evaluating the lateral hip in Greater Trochanteric Pain Syndrome (GTPS) relies on a systematic, anatomically precise, and dynamic approach. The patient is best examined in a lateral decubitus position with slight hip adduction and knee flexion, which relaxes the gluteal tendons and exposes the facets of the greater trochanter. A high-frequency linear probe (6–15 MHz) is used to scan the anterior, lateral, posterosuperior, and posterior facets, corresponding to the insertion sites of the gluteus minimus, gluteus medius (ventral and dorsal fibers), and gluteus maximus. For optimal visualization of the gluteus medius tendon, the probe should be oriented at a 45° angle to the femoral axis.
Dynamic maneuvers are essential. Using resisted motion tests such as FADIR (flexion–adduction–internal rotation), resisted abduction, and adduction allows the examiner to differentiate between gluteal tendinopathy, iliotibial band compression, and bursal irritation. The "gluteal cuff concept" divides the lateral hip into three layers—superficial (gluteus maximus, tensor fasciae latae, ITB), middle (gluteus medius, bursae, fat pads), and deep (gluteus minimus, short rotators)—which helps localize pathology accurately.
In most patients, GTPS represents gluteus medius or minimus tendinopathy or partial tear, while isolated bursitis is uncommon. High-resolution and dynamic ultrasound, rather than static imaging alone, provides the best diagnostic accuracy.
In summary, the best technique for sonographic evaluation of the lateral hip in GTPS is a multiplanar and dynamic examination of the greater trochanter's facets in lateral and supine positions, with functional provocation tests and angled (≈45°) probe alignment. This approach allows precise assessment of the gluteal cuff tendons and iliotibial band, recognizing that GTPS is primarily a tendinous rather than a bursal disorder.

MSUS Academy: Finally, do you have any advice for young colleagues in training using musculoskeletal ultrasound?

CM: First, stay curious and hands-on.
Second, know the anatomy and combine biomechanics, and clinical reasoning. Ultrasound should not be used as a stand-alone imaging tool; its true strength lies in linking what you see to the patient's symptoms and movement. Learn to scan dynamically, to reproduce pain (sonopalpation), and to correlate with functional testing.
Third, seek mentors and community. Teaching others later is the most effective way to consolidate your own skills.
Fourth, aim for quality, not speed. Resist the temptation to become a "button pusher." Build an internal standard of excellence early on; e.g. always do the whole region with all the structures – this way you repeat.
Finally, enjoy it: a "living anatomy lesson" every day.
You are your own project — growth begins the moment you take responsibility for it.

MSUS
Publication of the month

Link

Ultrasound diagnostics of the shoulder

Ultrasound is ideal for examining muscle quality and muscle structure in comparison to the healthy side. High-resolution musculoskeletal ultrasound of the shoulder is a validated and excellent supplement to clinical evaluation. However, good sonoanatomical knowledge is essential for recognizing and correctly classifying possible pathologies and assessing the prognosis.