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SAP – SAPHO: Synovitis, Acne, Pustulosis, Hyperostosis & Osteitis Syndrome

NIHR BioResource – Rare Diseases study project

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Summary

SAPHO (synovitis, acne, pustulosis, hyperostosis and osteitis) syndrome is a rare inflammatory bone disease that affects one in ten thousand individuals with peak onset between 20-50 years old. SAPHO syndrome typically affects the sterno-clavicular joint and anterior chest wall including the breast and collarbones. It can be associated with palmo-plantar pustulosis, severe acne and other skin conditions. Currently there is no diagnostic test or approved treatment for these patients and although the disease is not life threatening, it significantly affects quality-of-life for both the patient and their family.

SAPHO syndrome may be linked to another disease referred to as CNO or CRMO (chronic non-bacterial osteomyelitis / chronic recurrent multifocal osteomyelitis) that affects children and young adults. Both conditions have variable onsets and clinical features which makes studying them a particular challenge.

We aim to develop new treatments and ways of using existing treatments. To enable this, we have support from the NIHR BioResource to gather a cohort of SAPHO and CNO patients. These patients will help us to understand the genetic cause of these diseases and to unravel changes in cellular function. Patients will also facilitate studies into the natural history of the disease and improve our provision of care to both patients and their carers.

To improve people's quality of life, patient engagement is critical in this under-researched area. Consequently, in partnership with the NIHR we have also set up a patient advisory group. This opportunity will allow us to increase awareness of this condition and therefore improve the management of patients on a national basis.
Study website: www.impahcs.org

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Recruitment Criteria

Inclusion

- Clinical diagnosis of SAPHO (synovitis, acne, pustulosis, hyperostosis, osteitis)
- Clinical diagnosis of CNO (chronic non-bacterial osteomyelitis) or CRMO (chronic recurrent multi-focal osteomyelitis)
- Clinical diagnosis of sterno-costoclavicular hyperostosis, acne-associated spondyloarthropathy, pustulotic arthro-osteitis
- Joint or bone inflammation after retinoid therapy
- Unaffected relative (relative of a patient, where patient has a diagnosis listed above and no symptoms)
- Affected relative (relative has a diagnosis listed above with skin manifestations of palmo-plantar pustulosis, severe acne (e.g. nodulocystic acne) or hidradenitis suppurativa but no joint or bone features)

Exclusion

- Septic osteomyelitis
- Septic arthritis
- Diffuse idiopathic skeletal hyperostosis as a primary diagnosis
- Chronic polyarthritis such as rheumatoid or psoriatic arthritis as a primary diagnosis
- Ankylosing spondylitis as a primary diagnosis
- Benign or malignant bone tumours such as Ewing’s sarcoma