

JSC. Juvenile Systemic Sclerosis

NIHR BioResource – Rare Diseases study project

Lead Investigator: Dr Valentina Leone

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Summary

Juvenile systemic sclerosis (JSC) is a rare multisystem connective tissue disorder characterised by hardening skin changes and widespread internal organs involvement which presents before the age of 16 years.



Dr Valentina Leone, JSC project Lead

A UK study reported an annual incidence of 0.27 per million children and it is estimated that 3% of all patients with systemic sclerosis had onset in childhood. Similarly to the adult onset systemic sclerosis, there is a variety of clinical manifestations and disease course ranging from a slow to a rapid disease progression with early involvement of internal organs.

Children and young people with JSC show significantly less frequent involvement of internal organs at diagnosis including less common interstitial lung disease, pulmonary hypertension and renal involvement. Arthritis and muscle inflammation are more common in JSC. The distribution of antibodies associated with the condition is also different in JSC. Cardiopulmonary involvement albeit rare, remains the leading cause of mortality and morbidity in JSC.

The studies comparing children and adults with systemic sclerosis show that the distribution and pattern of organ-involvement (subset) is different with the diffuse cutaneous form (with more extensive skin thickening) and overlap form being more common in JSC. However, when adults with juvenile and adult onset systemic sclerosis are compared in adulthood, much fewer differences were observed.

Research and evidence based management of JSC is hampered by its rarity and the lack of outcome measures to use in research studies so currently the management of

this condition relies almost entirely on the use of evidence derived from studies undertaken in adults with systemic sclerosis.

Whilst there is an on-going international effort to collect clinical information on patients with JSC aiming to develop new classification criteria and outcome measures for clinical studies (International Inception cohort for Juvenile Systemic Sclerosis), the NIHR BioResource provides an opportunity to deepen our understanding of this rare condition by establishing a UK database including genetic biomarkers. Critically, this will facilitate future national and international collaboration with adult and paediatric colleagues with an interest in this rare condition and hopefully the development of joint clinical trials including adults and children.

Recruitment Criteria

Inclusion

For early/possible juvenile systemic sclerosis*

- Clinical diagnosis of Raynaud's phenomenon AND
- Nail fold capillaroscopy changes AND/OR presence of SSc specific antibodies (anti-centromere, anti-topoisomerase/sCL70, anti-RNA polymerase III)

For juvenile systemic sclerosis diagnosis

- Clinical diagnosis +/-
 - 2007 PRES/ACR/EULAR Provisional classification criteria for Juvenile Systemic sclerosis
 - 2013 EULAR ACR classification criteria

Exclusion

Rare paediatric conditions characterised by diffusely thickened skin as the cardinal feature including eosinophilic fasciitis, progeria, phenylketonuria or pan-sclerotic morphea should be ruled out as they may fulfil EULAR ACR 2013 adult classification criteria.

**The adult criteria for early systemic sclerosis have not been fully validated and may have limitations in children; however the inclusion of children with early features of systemic sclerosis is important, to understand their risk of progression into fully established juvenile systemic sclerosis.*