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SAD – Systemic Autoinflammatory Disorders (AOSD and uSAID)

NIHR BioResource – Rare Diseases study project

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Summary

Adult onset Stills disease (AOSD) and undifferentiated systemic autoinflammatory syndromes (uSAID) are a rare group of disorders which are characterised by unexplained fevers and inflammation which can affect various body systems. Diagnosis of these conditions can be difficult since the symptoms are not disease specific and we lack suitable diagnostic tests. This often leads to delay in diagnosis and access to correct treatment.

Dr Sinisa Savic, SAD project Lead

Some of the related autoinflammatory conditions are known to have a genetic cause, but in the case of AOSD and uSAID, the precise cause is unknown. The clinical features, laboratory parameters and responses of therapy of AOSD and uSAID show strong overlap and accordingly we plan to collect data on both of these simultaneously. We plan to investigate genetic, clinical, and laboratory measures of immune function from AOSD and uSAID patients, and use this information to determine which biological mechanisms are important in development of these conditions. Eventually we hope that this work will help us to develop more robust diagnostic tests and improve the choice of therapies for these conditions.

Recruitment Criteria

Inclusion

<u>For AOSD - Adult onset Still's Disease</u>
As defined by either Yamaguchi or Fautrel's criteria (requiring biological therapy anakinra or tocilizumab).

<u>For uSAID - Undifferentiated systemic autoinflammatory disorders</u>
Defined in both adult and paediatric patients as:

- Presentation with signs and symptoms of sterile inflammation
- Excluded infection, malignancy, rheumatic disease, primary immunodeficiency and monogenic SAID
- Currently on treatment with colchicine or biological therapies (e.g. anakinra, tocilizumab) that is showing clinical evidence of efficacy.