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100,000 Genomes Project—Rare Disease

Dermatology

Atopy

Example: Severe multi-system atopic disease with high IgE

- Adult but with onset in childhood
- Severe persistent eczema
- Recurrent/chronic S. Aureus infections
- Asthma
- High IgE levels >5000
- Molecular diagnosis not made previously

Ichthyosis

Example: Autosomal recessive Ichthyosis

- Neonates, infants, children and adults with a history of generalised red, dry, peeling skin at birth with a mode of inheritance consistent with autosomal recessive transmission
- Excludes: Ichthyosis vulgaris; STS-related ichthyosis; Syndromic ichthyosis, Keratinopathic ichthyosis; Acquired ichthyosis

Skin Adnexa disorders

Example: Familial cicatricial alopecia

- Family history of cicatricial alopecia in at least one first or second degree relative
- Consultant confirmed diagnosis

Autoimmune Skin disorders

Example: Generalised Pustular psorias

- Presence of primary , sterile, macroscopically visible epidermal pustules on non-acral skin.
- >1 episode of acute postulation
- Excludes those where pustules are limited to psoriatic plaques

Keratodermas

Example: Palmoplantar keratoderma and erthrokeratoderma

1+ of:

- Diffuse palmoplantar keratoderma
- Focal keratoderma with or without nail involvement
- Pachyonychia congenita phenotype (focal keratoderma with pain and dystrophic nails, oral leukokeratosis and or follicular hyperkeratoses/cysts).
- Punctate keratoderma
- Striate keratoderma alone or with wooly hair
- Keratoderma with deafness
- Unusual/unique rare keratodermas occurring alone or as part of syndromes.
- Erythrokeratoderma

Other Examples Include:

- **Neurocutaneous disorders**
- **Skin Fragility disorders**
- **Sun exposure related conditions**
- **Ectodermal dysplasias without a known gene mutation**

Please see the link below for full list of inclusion and exclusion criteria including necessary prior investigations

Rare Disease Eligibility

https://www.genomicsengland.co.uk/information-for-gmc-staff/rare-disease-documents/

For any questions please contact the
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