Website information RDCN-Mosaic Disorders March 2023

Introduction

Mosaic disorders are a group of rare disorders which are collectively not so rare. They are defined as a group by a common disease mechanism – they are caused by a genetic mutation (change) in a single cell in the developing baby during pregnancy. This leads to the baby being born with a mixture of normal cells (not carrying the mutation) and disease cells (carrying the mutation). It is this mixture which is referred to as mosaicism. This event can happen to any child, and importantly is not inherited from either parent.

The effects of the mutation depend on lots of different things. The most important ones are when it occurs and whereabouts it occurs in the body. In general, the earlier the mutation happens, the more serious the disease will be. As a result of the variability, each child with a mosaic disorder is unique in their disease and needs to be assessed uniquely. Mosaic disorders affecting the skin are currently the best known because it is possible to see the effects on the skin as birthmarks. As a result, although they often affect many body systems as well as the skin, mosaic disorders affecting the skin are looked after by Paediatric Dermatologists and Dermatologists. This RDCN will focus on the severe end of the spectrum of these diseases.

Aims of the RDCN-Mosaic Disorders

The overarching aims of the RDCN-MDs are in line with those for all RDCNs:

- to increase knowledge and understanding of mosaic disorders
- to progress research
- to improve patient experience.

The specific aims of the RDCN-MDs are

- to reduce mean time to first seeing a specialist
- to reduce the number of trips to the specialist centre
- to improve access to accurate clinical and genetic diagnosis
- to improve transition from paediatric to adult services, and to provide new adult access to specialist opinion
- to improve coordination of care between the RDCN and local hospitals

List of mosaic disorders which can be referred

Congenital naevi of all types

- melanocytic – small single lesions excluded
- epidermal – small single round sebaceous naevi excluded
- adnexal - any
- connective tissue – any

Vascular malformations of all types

- Arteriovenous – any
- Venous – small single asymptomatic lesions excluded
- Capillary – small single lesions excluded
- Lymphatic – any
- Mixed – any

Suspected mosaic disorders of unknown diagnosis – these would often be indicated by asymmetry of growth (overgrowth or undergrowth or body parts), extensive or multiple birthmarks (same type or multiple types), neurocutaneous disorders which don't fit with known germline diagnoses

Mosaic versions of germline diseases – for example mosaic neurofibromatosis type 1, mosaic CM-AVM syndrome, mosaic tuberous sclerosis, mosaic Darier disease, mosaic Gorlin syndrome

Specific exclusions

- Infantile haemangioma
- X-linked germline disorders which present with Blaschkolinear patterning such as incontinenti pigmenti, Goltz disease

Who can refer to the RDCN-MDs

Referrals can be made by a GP or a hospital consultant from any specialty. NB referrals cannot be made by patients themselves.

Referral criteria to RDCN-MDs

In the interest of expediting access to specialist services, referrals will be accepted from all UK GMC registered doctors including GPs, subject to the criteria below being fulfilled.

Mandatory primary RDCN-MDs referral criterion

Every patient referred into the RDCN service must have a clinician in charge of coordinating their care in their local hospital. This is to optimise coordination of patient care and communication between the RDCN and the local team, and to reduce the requirement for patient travel. This clinician must either be the referring clinician or be named in the referral letter if coming from a GP. Referral to the local clinician at the time of GP referral and naming of that clinician in the referral letter is sufficient at that stage, so that referral is not delayed by waiting for local appointments. This named clinician will be expected to remain involved in patient care throughout their care under the RDCN. For children the named clinician would be expected to be in Dermatology or Paediatrics, and for adults in Dermatology. However, for patients requiring systemic targeted therapies for their mosaic disorders (for example Sirolimus, Trametinib, Miransertib, Dabrafenib) an oncology professional may be the most appropriate even though these drugs are being used for mosaic disorders and not malignancy.

For a list of professionals who have already demonstrated an interest in being involved and are therefore open to referrals please see here:

Secondary RDCN-MDs referral criteria - to Great Ormond St Hospital

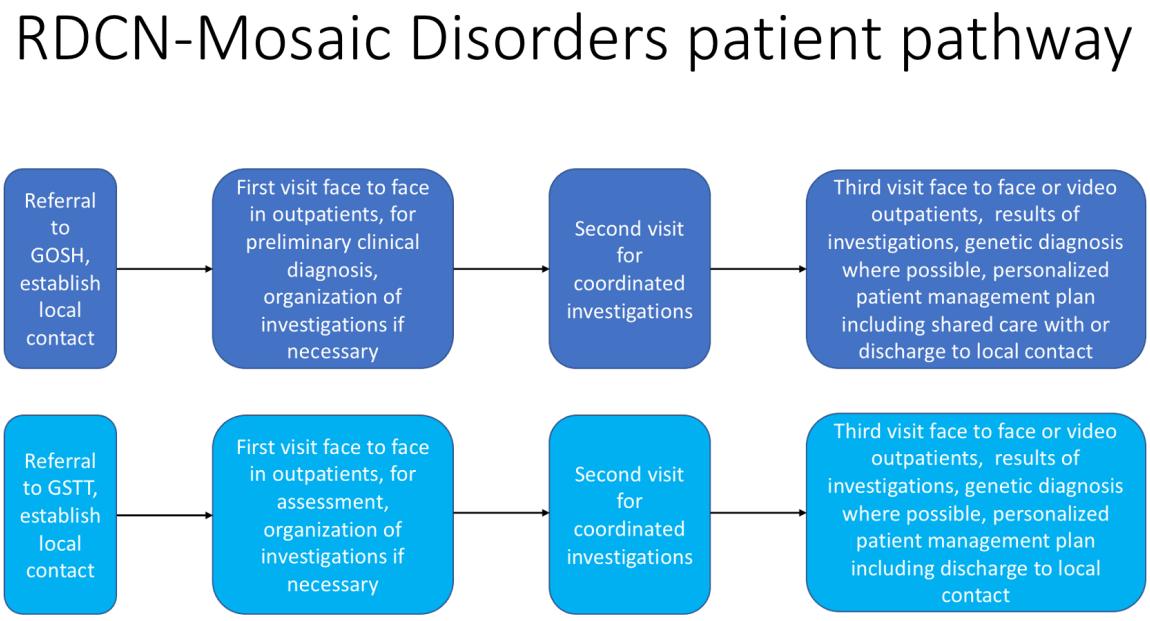
1) Suspected MD but no clinical diagnosis
2) Known MD but no genetic diagnosis
3) Suspected or known MD with clinical problems requiring management plan

Secondary RDCN-MDs referral criteria - to Guys and St Thomas' Hospital

1) Suspected MD significantly affecting life but no clinical diagnosis
2) Known MD but no genetic diagnosis and significantly affecting life
3) Suspected or known MD with new acute clinical problems

Patient pathway in the RDCN-Mosaic Disorders

Please note that this pathway is an outline and guideline only, and may vary between patients



Referral form for RDCN-Mosaic Disorders services

Mandatory primary criterion for GOSH and GSTT

Name of local hospital clinician in charge of care:
Local hospital name and address where that clinician is based:
Dropdown list

1) I am the local hospital clinician in charge of care
2) I have made a referral to the named clinician
3) The clinician has accepted the referral

Secondary criteria GOSH

1) Reason for referral
    1) Suspected MD but no clinical diagnosis
    2) Known MD but no genetic diagnosis
    3) Suspected or known MD with clinical problems requiring management plan
2) Diagnosis – drop down list (including unknown)

Secondary criteria GSTT

1) Reason for referral
    a. Transition from GOSH for continuing coordination of care from RDCN
    b. Suspected MD but no clinical diagnosis
    c. Known MD but no genetic diagnosis
    d. Suspected or known MD with clinical problems requiring management plan
2) Diagnosis – drop down list (including unknown)