

Appendix 6: Centre for Rare Diseases and ITM CRF



Centre for Rare Diseases (CfRD)

The Centre for Rare Diseases (CfRD) forms part of the Institute of Translational Medicine (ITM) and will be celebrating its fifth anniversary in September 2020.

There have been many developments, collaborations and achievements for the CfRD during this time including Biobanking, 100K genomics project, NIHR Bioresource (both Rare Diseases and Tissue Banking) as well as continual staff training for fibroscans and spirometry in order for these to take place as part of the patient pathway.

There has been the expansion of existing clinics to include different specialties to incorporate the “one stop” model, clinics increasing in frequency as well as being joined by colleagues from Birmingham Heartlands Hospital.

The CfRD is the location to 86 clinics (2 of which are virtual clinics) over 19 specialties. Activity has been steady with 10,409 appointments having taken place from April 2019 to January 2020, giving an overall total of 42,472 since opening in September 2015.

Discussions are continuing for the development of a bespoke Rare Diseases Registry for Birmingham to include the adult and paediatric centres. This will provide more accurate coding of the various rare disease cohorts; allow enhanced phenotyping and the potential to attract pharmaceutical company investment for the development of studies and new treatments.

NIHR Bioresource – Rare Diseases has recruited 247 patients to date. Neuroendocrine Tumours, Primary Sclerosing Cholangitis and Birdshot Chorioretinopathy are now included on the NIHR Bioresource portfolio and recruitment from these disease areas has commenced.

NIHR Bioresource - Tissue Bank (Healthy Volunteers) has gained momentum this year and there have been 105 recruits to this study to date. Recruitment includes the relatives of patients attending rare disease clinics plus members of Trust staff following an awareness of the study from the Communications bulletin.

604 patients have been recruited into rare disease studies in the time period April 19-January 2020.

In 2018/19 the first “virtual clinic” go live was achieved as part of a research study, for patients attending the Primary Sclerosing Cholangitis Clinic (PSC). The nationally

commissioned Bardet Biedl Clinic has recently undergone a test virtual clinic and this clinic is scheduled to go live in quarter 1.

Institute of Translational Medicine Clinical Research Facility (ITM CRF)

The ITM CRF, collocated with the CfRD, continues to attract phase III study delivery across a variety of specialities. These studies may be delivered solely in the ITM CRF by R&D teams i.e. Rubato (Cardiology) and Magnify (Neurology) or by R&D teams in collaboration with the NIHR CRF i.e. SUNRISE (Dermatology) and Monofer (Gastroenterology).

There are currently 25 live studies being delivered through the ITM CRF, a further 3 in collaboration with the NIHR CRF and 15 studies are at the set up stage.

193 patients have been recruited in to ITM CRF studies from April 19-January 2020