

## DC Action Draft Annual Report 2019

### Communication

Cheat Sheets (speciality-specific information suitable for quick reference by doctors/patients) now approved by Prof Sharon Savage and medical advisors and ready for website.

Medics for rare diseases (M4RD)- Instagram blog Oct 2019 featuring DC. Excellent uptake (>50 views which is well above average)

Personal contact with 3 new families

### Research

NCARDRS, patient experience, telomere length monitoring project proposal complete. Currently investigating funding sources. This will enable estimates of minimal prevalence of telomere biology disease (TBD) in the UK, understanding of the clinical features. *This is an essential prerequisite for developing appropriate NHS services and enabling trials of repurposed and new treatments.*

Contributed to Aplastic anaemia trust (AAT) NCARDRS project proposal (funded via King's).

British Lung Foundation grant co-applicant (with Exeter University)- early diagnosis of pulmonary fibrosis. *Early diagnosis will enable treatments to be tested at a potentially more reversible stage of the fibrosis process.*

Published arteriovenous malformations paper (with CCCTAA consortium: Higgs *et al.* Angiogenesis 22(1):95-102) *Raising awareness of this common and lifethreatening complication of TBD.*

DC Action members contributed to 2 online research surveys: i) Concord online survey study Genetic Alliance/UCL (resource requirement & co-ordinated care for patients with DC) and ii) patient attitudes to gene editing online study UCL/Warwick (on behalf of DC Action).

### Outreach

Contact with 3 more UK families

Attended UK and US patient meetings- informal contact and useful discussions with approx. 10 families and US physicians

### Clinic

Meetings re Cambridge SDS/DC clinic- no further progress

Possibility of standalone DC clinic under auspices of Cambridge genetics with Dr Anne Kelly (Paediatric haematologist)

### Contacts

Professor Graeme Alexander (Hepatologist) has joined our medical advisory panel (Jan 2019)

Inaugural members of 'Together for Healthy Marrow (TFHM). Consortium of charities supporting patients with bone marrow failures (Aplastic anaemia trust, Fanconi Hope, PNH support, DBA UK). Regular meetings since April 2019 and collaboration on research.

Dr Anna Duckworth PhD student in ILD genetics/diagnosis- collaborating on grant proposal and review paper (role of herpes viruses in telomere biology disease/ILD)

Dr Cham Herath. Astra Zeneca (AZ). Willingness to assist with NGS genetic diagnosis of at risk cohorts. Would need to identify cohort/collaborator and make written application.

### **Meetings**

ILD meeting (Nurses in interstitial lung disease) Birmingham, Oct 2019.

HL gave an oral presentation

Camp Sunshine patients' meeting (organised by Team Telomere) Sept

2019

Garstang family day (Team Telomere) July 2019

St George's Bone Marrow Failure meeting (patients, co-organised by AAT)

May 2019

King's Bone Marrow Failure meeting (Doctors/scientists) Oct 2019

CCCTAA (doctors) Autumn meeting Sept 2019

Findacure Drug Repurposing Meeting (Wendy) 27<sup>th</sup> Feb 2019

Name	Payment amount	Purpose
Hilary Longhurst	£126.00	DC action meeting
Jane Paxton	£67.15	Genetic Alliance meeting
Hilary Longhurst	£44.00	Findacure meeting
Hilary Longhurst	£126.00	DC Action meeting
Hilary Longhurst	£34.00	Camp Sunshine Dyskeratosis Congenita M
Hilary Longhurst	£90.57	Camp Sunshine Dyskeratosis Congenita M
Hilary Longhurst	£950.00	Camp Sunshine Dyskeratosis Congenita M
Hilary Longhurst	£10.00	DC Action stakeholders meeting, London
Total	£1,447.72	

Repayment required?	Date paid	Details
No - voluntary contribution	15-Jun-18	Mileage bath 280 miles
No	08-Jun-18	Train fares, subsistence
No	16-Aug-18	Mileage Lond 110 miles
No	23-Aug-18	Mileage Lond 280 miles
No	12-Sep-18	Train to airport
No	12-16 Sept 2018	Car hire excess (fuel, satnav etc)
No	12-16 Sept 2018	Flights, car hire
No	05-Oct-18	Refreshments (biscuits)