

MANTRAA : “MANchester Teaching and Research For RAre Disease Group

Annual Report

February 2021 to 2022

1. Introduction

MANTRAA was set up as a CIO in September 2019. The main objectives of the CIO is to advance and promote the science and medical practice of Rare Disease Genomic Medicine

To this end MANTRAA aims to:

1. Further education in Rare Genetic Diseases, supporting those who have an interest in this area to diagnose, understand, prevent, alleviate and cure congenital malformations and rare genetic disorders
2. Support rare disease meetings, specifically the Rare Disease International Meeting held in Manchester every two years
3. Support training through bursaries and scholarships
4. Aid the collation and dissemination of reports, statements and research findings

2. Changes to Trustees 2021-2022

Trustees remain the same but Sofia Douzgou has now relocated to Bergen, Norway , and Jill Clayton-Smith has retired from the NHS at the end of October 2021. Both have elected to remain as Trustees at present.

3. Work February 2021- February 2022

a) Meeting support

The instability following the COVID pandemic has meant that it has not been possible to risk planning an international Dysmorphology Meeting yet. It is deemed that the financial risk would be too great. However, things are starting to reopen and we are hopeful that a meeting can be planned for 2023. Traditionally this has always been in the autumn. Although we discussed the possibility of moving the meeting , it is likely that we will keep to autumn, avoiding half term and other major meetings. We need to make the decision as to whether to run this as a totally face to face meeting or a hybrid online and face to face meeting. In Feb 2022 a decision was made that the meeting would be held in Manchester from 15th to 17th October 2023.

b) Education

We have not put on any specific educational events under the MANTRRA umbrella as yet, again these have been limited due to COVID risk. We have contributed to other educational initiatives

c) Training

We were expecting to welcome a number of trainees to Manchester for training in rare diseases in May 2020. Unfortunately, given the COVID restrictions this had to be cancelled and the regulations for visitors have changed somewhat. We will plan to have trainees back in Manchester and possibly Bergen as soon as travel and risks allow. Due to the fact that trainees have not been able to travel, we have not given out any travel bursaries or scholarships yet.

d) Collation and dissemination of reports

We have contributed to compilation of some reports on management of rare diseases, though have not yet published any under the MANTRRA umbrella. In particular we have been working on management guidelines for Kabuki Syndrome and PIK3CA related overgrowth syndromes where we have linked with parent support groups. One of the trustees continues to act as an editor for a Rare Disease Journal (Clinical Dysmorphology) with the honorarium for this coming into the MANTRRA funds.

4. Impact of Covid 21-22

As outlined above, we have been severely impacted by COVID, with the requirement to rearrange our conference and defer trainees. We were also due to have been involved in hosting a workshop at the European Society for Human Genetics in June 2020 and this was cancelled. We are now planning to do this as a virtual session at the next meeting in June 2021. The fact that no-one is travelling to meetings at present means that we haven't given out any travel bursaries.

5. Impact of Brexit

By virtue of the fact that the conditions MANTRRA is aligned to are rare, specialists are few and far between and dialogue and communication between experts nationally and internationally is paramount. Our network is world-wide, but we have greater interactions with our European Colleagues. Our team used to be involved in the coordination of ERN ITHACA, one of the European networks for rare diseases. Unfortunately, as we came up to leaving the EU we had to stand down as coordinators and can now no longer be members. This has some impact on our ability to work together but in fact we have well established relationships with our EU colleagues and at the present time there is no significant impact for us, although in future we may offer funding via MANTRRA for education/training /research activities as funding will not be available from the ERN.

6. Trustee/Admin Changes

One of the MANTRRA trainees, Sofia Douzgou has now moved to live and work outside the UK in Norway but remains a trustee. This means that trustee meetings and decisions now need to be virtual or by electronic communication. It has the advantage that Dr Douzgou remains within the EU and an active participant in the European Reference Networks, so maintaining our contacts.

7. Future Plans for 2022-2023 (contingent on COVID situation)

1. Reorganisation of Manchester Dysmorphology Conference (October 2023)
2. Consider other ways of trainee support if bursaries/scholarships cannot be taken advantage of at the present time
3. Continue management guideline work and participation in other virtual educational initiatives.

8. Accounts

Our income comes mainly from Honoraria for work from the Clinical Dysmorphology Journal, from other honoraria and from occasional donations. We had some initial commercial funding from an exhibitor at a meeting held in 2018.

Our main initial aim has been to accumulate a reserve fund for conferences, then look further at providing funds for other aspects of training/research. In the event we have not spent any of our funds as yet given the reasons above.

Date	Item		Receipt	Expenditure
April 2021	Opening balance	28,172.05		
January 2022	Journal honorarium		3,550	
	Closing balance	31,72205		

BALANCE £31,72205 as of 1st April 2022

Annual income 21-22 £3550

Report reviewed and finalised March 2023 by Jill Clayton-Smith