

# **MANTRAA : “MANchester Teaching and Research For RAre Disease Group**

## **Annual Report**

1/4/20 to 1/4/21

### **1. Introduction**

MANTRAA was set up as a CIO in September 2019. The main objectives of the CIO are 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. Work since inception**

#### **a) Meeting support**

Initial work of the three trustees of MANTRRA ( Douzgou, Banka, Clayton-Smith) focussed on preparation for the 2020 Manchester Dysmorphology Conference , due to be held in November 2020. Various venues were researched and visited to identify suitable conference accommodation for around 220 participants, with an emphasis on encouraging participation from as many trainees as possible. Eventually, The Kimpton Hotel, Manchester was identified as the most suitable venue and a contract negotiated and signed with the hotel in early March 2020.. A search was instigated for administrative help with the conference.

By the spring of 2020, as the COVID pandemic hit , it was clear that there would be too much risk for a November 2020 conference to go ahead and after negotiation with the conference venue the date was deferred until May 2021 and then until November 2021. Currently, with the second wave of the pandemic and recent introduction of travel restrictions to the UK, a deferral of the conference until at least November 2022 was envisaged , as the aim was to have an international audience. All of these rearrangements have given rise to many discussions among the trustees. As of 1/4/21 we planned provisionally for a 2022 meeting, and were also possibly considering alternatives such as a virtual meeting. A decision was made to look into the cost of this.

#### **b) Education**

We have not run any specific educational events under the MANTRRA umbrella by April 2021, again as these were limited due to COVID risk and redeployment of doctors within the NHS to cope with the COVID pandemic. We contributed to other educational initiatives , for example an on-line course which is being developed by the European Reference Network for Congenital Malformations and Intellectual Disability.

### c) Training

We were expecting to welcome a trainee from Slovenia to Manchester for training in rare diseases in May 2020. Unfortunately, given the COVID restrictions this had to be cancelled. We plan to have trainees back in Manchester 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. All of the trustees did, however, contribute to training activities outside their NHS work by lecturing at various meetings. We also contributed to the setting up of the European Society of Human Genetics ESHG Young group for trainees in all aspects of genomics.

### d) Collation and dissemination of reports

We 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.

## 3. Impact of Covid

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.

## 4. 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.

## 5. 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.

## 6. Future Plans for 2021-2022 (contingent on COVID situation)

1. Reorganisation of Manchester Dysmorphology Conference

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.

## 7. 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.

Accounts for April 2020 to April 2021

| Date      | Item            | Amount             | Receipt | Expenditure |
|-----------|-----------------|--------------------|---------|-------------|
| 1/4/2020  | Opening Balance | 21,072.05          |         |             |
| 10/7/2020 |                 | Journal honorarium | 3550.00 |             |
| 29/1/2021 |                 | Journal honorarium | 3550.00 |             |
|           | Totals          |                    | 7000.00 |             |
| 1/4/2021  | Closing balance | 28,172.05          |         |             |

**BALANCE 28,172.05 as of 1<sup>st</sup> April 2021**