

MANTRRA: “MANchester Teaching and Research for RAre Disease

Annual Report

April 6th, 2024

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:

- a. 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
- b. Support rare disease meetings, specifically the biannual Manchester Dysmorphology Conference. The first one was organized in 1984 by Professor Dian Donnai. Over the years, this biennial conference has brought experts within the field of genomic syndromology together from around the globe, to share knowledge and ideas which further our understanding of developmental disorders and congenital malformations. The conference aims to provide cutting edge content and a welcoming, relaxed setting to encourage discussions, forge collaborations and inspire new ways of improving the lives of patients.
- c. Support training through bursaries and scholarships
- d. Aid the collation and dissemination of reports, statements and research findings

2. Changes to Trustees 2023-2024

No changes to Trustees. Sofia Douzgou Houge (SDH) is resident in Bergen, Norway. Ongoing discussions regarding the appointment of further trustees (invited but not yet accepted).

3. Activities February 2022- February 2023

- a. **AGM.** Our AGM was held on October 17th, 2023.
- b. **Annual reports.** SDH circulated a 2023-2024 report late and will submit asap.

4. Meeting support

The 2023 Manchester Dysmorphology Conference: From October 16th to October 18th, 2023, the Hilton Deansgate in Manchester hosted 252 international genomics experts, fostering knowledge-sharing and idea exchange. This conference advanced our understanding of developmental disorders and congenital malformations. With 83 cutting-edge presentations on the science, diagnosis, and treatment of rare conditions, it not only delivered top-notch content but also fostered a welcoming and relaxed atmosphere, facilitating discussions, promoting collaborations, and inspiring innovative approaches to enhance the well-being of patients affected by rare conditions. Participants hailed from 50 countries worldwide.

We agreed to provide expenses for building the previous conference website to Cristian Cunha. A fee of £2000 was paid. A website is important for a large meeting and also helped with marking and collation of abstracts.

5. Education

We continue to contribute to other educational initiatives through our links with the European Reference Network for Congenital Anomalies, ERN ITHACA and the European Society of Human genetics (ESHG). SB and SDH have continued to lead and participate in teaching activities in the UK and overseas. Prizes for presentations were awarded at the Manchester Dysmorphology Conference also supported by ERN ITHACA. MANTRAA supported educational activities organised by Dr Grace Vassallo for one of the commonest rare genetic condition called Neurofibromatosis 1 (NF1). Individuals with these disorders need specialist care and it's important to train GPs and other doctors to both recognise signs of these disorders and to be able to help with specialist management.

6. Collation and dissemination of reports

We continue to contribute to compilation of publications for rare conditions in the medical literature. We reiterated that we need to mention MANTRRA in the acknowledgements when this type of paper is published.

7. Future plans

- a. Organization of the ESHG Manchester Syndromology & Dysmorphology Course from 22nd – 24th October 2024
- b. Organisation of the 20th Manchester Dysmorphology Conference 16th-19th Nov 2025
- c. Continue with meeting support. Foster new liaison with NF1/NF2 group
- d. Continue management guideline work and participation in other virtual educational initiatives.

8. Accounts

Date	Item		Receipt	Expenditure
06/04/2023	Starting balance	£41,974.01		
27/11/2023	NF1 educational meeting organisation			-£312.31
Totals			0	-£312.31

BALANCE £41,661.70 as on 6th April 2024

Report compiled August 2024 by Sofia Douzgou Houge, Siddharth Banka and Jill Clayton Smith