

MANTRRA: “MANchester Teaching and Research for RAre Disease

Annual Report

March 9th 2025

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 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.
3. Support training through bursaries and scholarships
4. Aid the collation and dissemination of reports, statements and research findings

2. Changes to Trustees 2024-2025

Jill Clayton-Smith ended her membership. Sofia Douzgou Houge (SDH) is resident in Bergen, Norway. All trustees agree to close the charity within the following financial year.

3. Activities February 2024- February 2025

- a) **AGM.** Because of a busy schedule around point 3c), and on-going discussion around closing the charity (see point 2), the AGM took place over mail correspondence.
- b) **Annual reports.** Trustee members received notification about a delayed annual report on Friday 7th March 2025, SDH circulated the 2024-2025 report on March 9th, 2025, and will submit asap.
- c) **Meeting support**

The 2024 Manchester European Society of Human Genetics (ESHG) Syndromology and Dysmorphology Course: the course was held in Manchester, October 22nd- 24th, 2024. It was advertised on the ESHG website, the Manchester Rare Conditions Centre (MRCC) website and on relevant social media platforms. We reviewed 134 applications and 74 national, and international (Manchester to Hong Kong) attendees were offered participation. Speakers (18) from countries across the European continent were invited. Feedback from attendees was very positive with an average rating around overall organization of the course 4.79/5.

We have had some discussions with Dr Grace Vassallo and Dr Gareth Evans who hosted the Euro NF1 and NF2 conference in 2022. Neurofibromatosis 1 and 2 are multisystem disorders, with NF2 being very rare. Individuals with these disorders need specialist care and its important to train GPs and other doctors to both recognise signs of these disorders and to be able to help with specialist management. We have agreed to support educational initiatives in the future, particularly training expenses for doctors.

d) Education

We continue to contribute to other educational initiatives through our links with the European Reference Network for Congenital Anomalies, ERN ITHACA. SB and SD have continued to participate in teaching activities in Europe, the UK and overseas. ERN ITHACA sponsored fellowships for the ESHG and MRCC course too.

e) Collation and dissemination of reports, statements and research findings

We continue to contribute to publications for rare conditions in the scientific medical literature.

4. 2025-2026 Plans

- a) Scientific support of the Manchester Dysmorphology Conference November 2025
- b) Continue with meeting support liaison for the Manchester NF1/NF2 group
- c) Closure of the charity

5. Accounts

These were verified over email.

We declare no further income this year.

We will have to pay an accountant for audit as per the Charity commission rules. That charge will be added to our account later.

Our expenditure was for support for NF team (see point 3c).

Date	Item		Receipt	Expenditure
February 2024	Starting balance	£39,194.05 of which £9,472.00 donation from Convenzis Group (NF conference net income)		
November 2023	Transfer to Dr G Vassallo for NF1 MDT expenses	-£312,31		

April 2024	Transfer to Dr G Vassallo for NF1 MDT expenses	-£269,90		
November 2024	Transfer to Dr G Vassallo for NF1 MDT expenses	-£299,35		
Totals		£38,312.44 of which £8,590.44 NF fund		

BALANCE £38,312.44 as of February 2025

Report compiled March 2025 by Sofia Douzgou Houge and Siddharth Banka