Mahmoud Koko

Dr. Mahmoud Eltayeb Koko Musa
Cambridge, UK
Mobile: 0044 7523 6343 80

Email: mahmoudkoko@outlook.com
Portfolio: mahmoudkoko.github.io
LinkedIn: linkedin.com/in/mahmoudkoko
ORCID: orcid.org/0000-0001-9512-0184

Experienced genetic researcher and genomic analyst with a strong background in molecular medicine, neurogenetics and computational genomics. Specialised in sequencing data analysis at scale. Passionate about leveraging large-scale data to drive translational insights in medical and population genetics. Invested in roles that bridge research and clinical application in genomics.

EDUCATION AND QUALIFICATIONS

- 01/2023: Doctorate in Neuroscience (*summa cum laude*), University of Tübingen, Germany.
- 02/2017: Registered doctor (no. 7361103), General Medical Council, United Kingdom.
- 11/2014: Master's degree in Molecular Medicine, University of Khartoum, Sudan.
- 12/2013: Registered doctor (no. 41118), Sudan Medical Council, Sudan.
- 01/2012: Bachelor of Medicine and Surgery, University of Khartoum, Sudan.

RESEARCH EXPERIENCE

- 10/2022 – 10/2025: Postdoctoral Fellow, Wellcome Sanger Institute, Hinxton, UK.
  - Studied the role of rare variants in sex differences in autism prevalence in ~46,000 autistic individuals from ASC and SPARK cohorts.
  - Contributed to curation of ~37,000 exomes from UK birth cohorts.
  - Investigating the relationship between rare non-coding variants and cognition in ~500,000 UK Biobank genomes.

- 10/2022 – 10/2025: Visiting Research Associate, University of Cambridge, UK.
  - Part of the Prenatal and sex differences in autism consortium (APEX) at the Autism Research Centre (Department of Psychiatry).
  - Studying the genetic basis of predisposition and heterogeneity in autism using clinically ascertained and birth cohorts.

- 10/2016 – 11/2021: Doctoral Researcher, Hertie Institute, Tübingen, Germany.
  - Analysed exome sequencing data from ~10,000 individuals with epilepsy from several consortia (Epi25, EuroEPINOMICS-CoGIE, EpiPGX, CENet, Epi4K, EP/GP).
  - Used computational models and functional evaluation to inform diagnosis and personalised therapies in disorders of ion channels and transporters (e.g., *SCNxA*, *GABRx*, *KCNxx*.)

- 01/2014 – 09/2022: Collaborating Research Fellow, University of Khartoum, Sudan.
  - Participated in establishing the Sudanese neurogenetics research group ("NeuroGeneticsSudan").
  - Investigated novel variants and genes underlying neurological diseases in consanguineous Sudanese families (spinocerebellar degeneration, movement disorders, leukodystrophy, muscular dystrophy, epilepsy).
  - Contributed to studies of population genetics and cancer genetics in the Sudanese population.

SKILL SET

- Statistical, computational and functional analysis of ultra-rare variants.
- Sequencing data QC and analysis at scale.
- Data exploration, analysis and visualization.
- Coding, code documentation and management (git, containers).
- Using Linux HPC clusters and cloud computing environments.
- Project leadership, academic supervision, writing and presentations.

A1	C2	Lang.
★★★★★★		Arabic
★★★★★☆		English
★★★★☆☆		German

TEACHING APPOINTMENTS

- 05/2015 – 05/2016: Fellow Lecturer, University of Khartoum, Sudan.
    - Lecturer at the Institute of Endemic Diseases (IEND).
    - Taught graduate courses in Introductory Bioinformatics and Molecular Biology
    - Organized and delivered workshops in genomics and bioinformatics.

- 01/2015 – 05/2016: Clinical Tutor, Ahfad University for Women, Sudan.
    - Tutor at the Department of Clinical Skills and Problem Based Learning (PBL), Ahfad School of Medicine.
    - Taught undergraduate practical courses in bedside clinical skills and moderated PBL sessions.

CLINICAL APPOINTMENTS

- 03/2014 – 11/2014: Doctor, National Service Administration, Sudan.
    - Medical Officer taking primary responsibility for non-specialist in-patient and outpatient care.
    - Worked in clinics affiliated with Ribat University Hospital (March - July) and Omdurman Military Hospital (August - November).

- 10/2012 – 10/2013: Doctor, Federal Ministry of Health, Sudan.
    - House Officer (internship doctor) providing in-patient, out-patient and emergency care.
    - Did rotations in pediatrics, surgery, obstetrics & gynecology and internal medicine.

- 02/2012 – 07/2012: Doctor, National Service Administration, Sudan.
    - House Officer (internship doctor) providing A/E, in-patient and out-patient care.
    - Worked at Khartoum E.N.T. Hospital (February - April) and Ribat University Hospital (June - July).

MEMBERSHIPS AND AWARDS

- Member of the European Society of Human Genetics (no. 11455).
- Tuebingen University Clinic (UKT) scholarship (2021).
- German Academic Exchange Service (DAAD) doctoral award (2016 – 2020).

SELECTED PUBLICATIONS

- Koko *et al.* (2025). Contribution of autosomal rare and *de novo* variants to sex differences in autism. ***AJHG***. PMID: 39954678.
- Koko, Fabian, *et al*. (2024). Exome sequencing of UK birth cohorts. ***Wellcome Open Research***. PMID: 39839975.
- Koko *et al*. (2024). Bi-allelic PRRT2 variants may predispose to Self-limited Familial Infantile Epilepsy. ***EJHG***. PMID: 38316952.
- Koko *et al*. (2022). Association of ultra-rare coding variants with genetic generalized epilepsy: A case–control whole exome sequencing study. ***Epilepsia***. PMID: 35032048.
- Koko *et al*. (2021). Distinct gene-set burden patterns underlie common generalized and focal epilepsies. ***EBioMedicine***. PMID: 34571366.
- Koko *et al*. (2018). Challenges imposed by minor reference alleles on the identification and reporting of clinical variants from exome data. ***BMC Genomics***. PMID: 29334895.

ORCID Profile  FULL PUBLICATION RECORD  Google Scholar

REFERENCES Available upon request