



REPUBLIC OF ZAMBIA  
MINISTRY OF HEALTH  
University Teaching Hospitals  
**CHILDREN'S HOSPITAL**

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OFFICE OF THE SENIOR MEDICAL SUPERINTENDENT

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**Our Ref:**

8<sup>th</sup> June 2018

**Your Ref:** Dear Professor Hanna

**INTERNATIONAL CENTRE FOR GENOMIC MEDICINE IN NEUROMUSCULAR DISEASES ZAMBIA**

**We confirm our participation in this new MRC strategic initiative if awarded according to the following principles with which we jointly agree:**

**Benefits for research and for the health of NMD patients**

We confirm that an International Centre for Genomic Medicine in Neuromuscular Diseases would have very significant and long lasting research and health benefits for our patients with NMD.

Defining our patient cohorts by genotype will allow us to:

- Increase the number of patients with a precise genetic diagnosis the results of which will be conveyed to patients and their parents/guardians, according to the local arrangements, through clinical services we lead
- Discover new genes
- Provide more accurate advice and counselling to patients and families
- Adapt as appropriate for our setting and implement international care guidelines for screening/managing patients on a rational basis
- Make the case locally for access to appropriate screening and simple health interventions including cardiorespiratory, gastroenterological, metabolic and physiotherapy.
- Identify patients who will benefit from using re-purposed cheap drugs for some conditions such as channelopathies and congenital myasthenias based on a precise genetic diagnosis

**Long lasting benefits of training and bioinformatics platform:**

- **Training a new generation of clinical academics** with strong UK support networks who will become African leaders of genomic medicine and specialist care delivery in and who will develop national networks of excellence and be a transformational output from this initiative
- **Sharing genetic data:** contingent upon ethics approvals, the openly shared bioinformatics data platform will be a major advantage this consortium and will

empower all clinicians and clinical researchers to analyse their own patients' NGS data on the RD-Connect genome phenome analysis platform and derive molecular diagnoses. They will continue to do so beyond the funding period of the centre. Moreover, they will be able to educate and train other healthcare professionals in their countries to make use of NGS data and bioinformatic platforms for the benefit of their patients.

These developments will only happen through this new initiative and will add major value to our existing developing neuromuscular services enabling us to become a centre of excellence which will spread good practice nationally and enable sustainability after 5 years.

### **Building Cohorts**

We assess over 14,000-15,000 paediatric inpatients per year with NM diseases being one of the commoner neurologic disorders in our setting. The outpatient clinics are even more extensive given that our Children's Hospital serves as the catchment area for the entire country of 15 million. With the support of clinical fellows, we will aim to recruit, phenotype and genotype at least 500 patients into the cohort over the next 5 years. As discussed, funding will be provided to develop systems to facilitate referrals from District Hospitals and/or mobile NMD teams to evaluate and track patients at the District level. We understand that recruiting the cohort is an essential deliverable for our centre and that it will be reviewed monthly.

### **Appointing fellows**

There will be major competition for such prestigious fellowships. We will undertake to advertise and appoint such fellows in conjunction with the training and fellowship committee chaired by Professor Reilly during the 6 month period prior to the grant initiation. We understand that appointing fellows is an essential deliverable of the programme. We will ensure fellows have two local mentors (a clinical mentor and a science mentor). We will minimise the risk of fellows moving abroad after training by working with the Ministries of Health and Education to craft appropriate positions within Zambia for them at the end of the fellowship to secure a permanent legacy of specialist clinical academics in neuromuscular genomic medicine for patients. Furthermore we note that during the two 6 month training periods in the UK they will not have full GMC registration but will only have observer status.

### **Delivering healthcare solutions**

The University Teaching Hospitals are the Academic Centres for Zambia with a burgeoning post graduate training program in Neurology--the only one in the country and one of the very few in sub-Saharan Africa. We confirm increasing the number of patient with a precise genetic diagnosis will be a significant healthcare benefit in our centre and we will be in a position to implement simple standards of care that are required following a precise genetic diagnosis. Furthermore it will allow us to build genotyped trial "ready cohorts" which we can follow up. For precisely defined genotyped cohorts where new treatments become available as is likely over the next 5-10 years we will seek to partner with governmental bodies and healthcare funders to make drugs available to our NMD patients.

### **Data sharing and ethics approvals**

All data will be pseudoanonymized. Contingent upon local regulatory approvals, we are fully committed to data sharing through the proposed platform. We recognise that data sharing is important to the project and will be made possible following an agreed embargo period and under controlled access conditions. We recognise that all data will adopt the General Data Protection Regulations of the EC 2018. We confirm we will secure all required local ethical approvals before MRC funding commencement.

### **Sustainability following the end of the grant award**

The initial investment from the MRC in this initiative will be a major catalyst for our centre to secure additional outside investment and ensure sustainability. By being part of this

application we have already been able to secure onward support for the fellows at the end of the grant to ensure they are a lasting legacy of neuromuscular expertise.

Our participation in this programme will enable us to leverage institutional and national funding schemes. We confirm we agree to all the above principles.

Yours sincerely

A handwritten signature in black ink, appearing to be 'M. Mwenechanya', enclosed in a large, loopy oval shape.

**Dr M. Mwenechanya  
A/SENIOR MEDICAL SUPERINTENDENT  
UTHs -CHILDREN' S HOSPITAL**

A handwritten signature in black ink, appearing to be 'G. Birbeck', written in a cursive style.

**Gretchen L. Birbeck, MD    Principal, UTHs Neurology Research Office**