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Application of whole-exome sequencing to identify novel disease causing genes associated with mitochondrial disease

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Advances in next generation sequencing technology have resulted in a rapid increase in the molecular characterization of mitochondrial disease. Recent years, our laboratory has successfully used whole-exome sequencing to identify many novel disease causing genes associated with mitochondrial disease. The mitochondrial asparaginyl-tRNA synthetase (*NARS2*) mutations cause Leigh syndrome and non-syndromic hearing loss (DFNB94). We found that some mutation can disrupt dimerization of NARS2 and decrease steady-state levels of mt-tRNA^{Asn} without aminoacylation defects. The cells with *NARS2* mutations also display impaired oxygen consumption rate and OXPHOS deficiency that can be rescued by overexpression of wild type *NARS2*. Recently, we found that recessive SLC25A46 mutations cause optic nerve atrophy and axonal peripheral neuropathy. SLC25A46, putative mitochondrial carrier gene, is human homologs of Ugo1p. Furthermore, we demonstrate the SLC25A46 role in mediating mitochondrial morphology *in vitro* and *in vivo*. In zebrafish, we found that loss-of-function affects the development and maintenance of neuronal processes and causes abnormal mitochondrial fusion morphology. Our result show many disease causing genes associated with mitochondrial disease are yet to be identified and whole-exome sequencing is very cost-effective for this process.

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The impact of care giving a child with HIV/AIDS: Experiences of Zimbabwean mothers

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Background & Aims: Zimbabwe is amongst the top nations affected by the HIV/AIDS worldwide. Literature posits that care giving a child with long term health condition may lead to deterioration of the health and HRQoL of informal caregivers. Further, the burden of care giving may be more if caregivers are of low socio-economic status and when the caregivers is also suffering from a chronic condition. The aim of the study was to determine the impact of care giving a child with HIV/AIDS and to compare with that of caregivers of children with cancer, CP and who were typically developing.

Methods: A cross sectional study was done at Parirenyatwa Central Hospital in Harare, Zimbabwe (MRCZ/B747). Caregiver burden and HRQoL were assessed using the caregiver strain index (CSI) and EQ-5D respectively on 108 primary, informal caregivers.

Results: Most of the children were males, n=55 (50.9%) with mean age of 28.5 months (SD13.5). All the caregivers were female and most were unemployed, n=74 (68.5%). Caregivers of children with HIV/AIDS reported of a higher burden of care, F (6, 206)=10.17, p<0.001 and reported of a poorer HRQoL, F (3,104)=11.81, p<0.001.

Conclusions: Informal caregivers of children living with HIV/AIDS are more likely to report of caregiver burden and poorer HRQoL. Although causality cannot be inferred, findings from the study suggest a great need for routine screening and formulating tailor-made interventions to alleviate the burden of care and improve HRQoL of caregivers of children with HIV/AIDS.

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