EDITORIAL

MEDICAL GENETICS AND EDUCATION

The recent dramatic cures announced for diseases like sickle cell disease, Spinal Muscular atrophy, Beta Thalassemia using revolutionary gene editing techniques and gene therapies, the availability of advanced genetic tests, raises the question of what currently goes into our medical training regarding medical genetics and genomics education. The human genome project was completed in 2005 that confirmed humans had only between 20000 and 23,000 gene pairs and that mutations in our genomes caused about 7000 known genetic diseases that affects millions of families worldwide. Ghana is no exception and at the Department of Child Health and now armed with access to whole genome sequencing investigations in the USA (Through Rare diseases Ghana Initiative) we are beginning to diagnose a whole new range of diseases hitherto thought non-existent in our environment. Rare diseases like Rubenstein- Taybi syndrome, Wilson's disease, SCN1A mutation epilepsy (Dravet syndrome), Gauchers disease to name a few are being seen in the Department. It has already been observed at the Neuro- Developmental clinic for instance that our children tolerate much lower doses for epilepsy drugs than what is prescribed in the books. The issue of pharmacogenetics comes in here where the variation in drug metabolizing enzymes could be a factor. In 1892 Sir William Osler said, "if it were not for the great variability among individuals, medicine might as well be a science and not an art'.

The brilliant gene editing techniques and known as CRISPR Cas9 or Clustered regulatory Interspaced Palindromic repeats associated protein 9 which acts as a' genetic tailor' has moved rapidly from being a laboratory technique to the clinical bedside. We depend on our survival as humans on our ability to repair mutations. Xeroderma pigmentosum is an example of such a failure. Mutations are the driving force of nature. Imagine copying a book such as the bible or the Koran as happens in DNA replication. Monks used to do that job. There were huge errors. This tool marks a new revolution in medical practice to treat genetic diseases.

There are references to genetic diseases and the diagnosis in a few articles. Is our clinical workforce trained to handle this major shift going on? I envisage laboratory reports dotted with genetic language in the near future. There is the issue of having your private genetic biodata available to insurance companies and the whole world. What are the undergraduate and post graduate institutions doing about these rapid developments regarding basic training in genetics? Bennet et al suggest we need to foster excitement about medical genetics specialties early upon entry to medical school as well as throughout their training. The content of genetic training should be looked at by our Curriculum review committees serving the medical schools. Genetic counselling training has begun at the West African Genetic Medicine Centre(WAGMC) , Legon and it is hoped our population will be served with their expertise. The public deserves to be informed by health professionals about the nature of disease and the role of genes. There is no surveillance and registry for rare genetic diseases and this should be quickly established so we can participate in International trials for new drugs.

References

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