DIVISION OF

HUMAN GENETICS

Maximising the benefits of the genomic revolution for Africa

Professor Raj Ramesar
Head of
Human Genetics

In the Division of Human Genetics there is never a dull moment. The vast diversity of genetic disorders, coupled with the explosion of knowledge about genetics and molecular biology means that the department's clinical and research teams are constantly finding new information about the conditions seen in their patients and their families.

"Being exposed to complex patients and working with inspirational colleagues across disciplines, including research technology platforms, leads to exciting and meaningful research, which directly translates into patient benefits," says Professor Raj Ramesar, who heads up the division.

"Planning tomorrow's research, of direct relevance to our clinics and patients, with intelligent and driven postgraduates, is possibly the most exciting thing we do."

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The past decade has seen a renaissance of sorts within the division. Since its inception, it has had a proud history as an extremely productive unit, both academically and clinically. However, for various reasons, by the turn of the century, the division's clinical component had been reduced to a few hardworking individuals and its continuation as a training unit was threatened.

Fortunately, the last decade has seen a steady growth and strengthening of the clinical team, along with the establishment of an efficient and innovative diagnostic laboratory and wide-reaching, productive translational research endeavors.

From a single doctor and two nursing sisters a decade ago in 2014, the clinical team includes two medical genetic specialists, three medical genetic registrars, two genetic nursing sisters and two genetic counsellors.

The past decade has also had some significant academic and training advances including the inception of the Master's programme in genetic counselling, the move of medical genetics from a subspecialty to a primary speciality, the commencement of regular Medical Genetic Education Programme (MGEP) courses for nurses, and, most recently, the addition of short courses in genetic counselling.

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Academic outputs, as measured by publications and postgraduate student supervision from the clinical team, have also shown an increase, due to an increasing recognition of the value of research and its translation into clinical practice.

Research in the division's laboratories has also found its way to the diagnostic laboratories of the National Health Laboratory Services, resulting in improved molecular testing, allowing more diagnoses to be made and more options for families across South Africa.

Administration has become more objective and better organised, with regular clinical meetings, improved diagnostic laboratory liaison and formal registrar and student assessments.

The division's growing stature bore fruit in 2011, when it organised and hosted a Joint International Conference of the African and Southern African Societies of Human Genetics. The event drew a record number of more than 500 delegates, mostly from across Africa.

This meeting, together with the Human Heredity and Health (or H3) Africa initiative, saw Africa being taken seriously by the international genetic community. The funding (by the National Institutes of Health in the USA, and the Wellcome Trust in the UK), for the initiation of the H3Africa consortium of projects, spanning Africa across a wide range of disease conditions, is certainly a dream come true, and one which will move towards improving the health of the citizens of our continent.

Genetics has massive potential to advance African health. Professor Ramesar says that in the years ahead, the division will seek to continue to improve its clinical service, informed by appropriate translational research, and drive cost-effective laboratory testing to improve the care it can offer to patients and their families.

Excellence in training for the next generation of medical geneticists – for South Africa and Africa – will also be a focus along with the education of communities and healthcare, workers alike and facilitating knowledge transfer between universities and the basic education sector.

"We want to allow maximum advantage to be taken of the genomic revolution in Africa, with careful attention to clinical utility and ethical practice," concludes Professor Ramesar.