As many as 60,000 South African babies born annually - and their families - may be in need of urgent assistance in dealing with serious genetic disorders and birth defects.

Professor Arnold Christianson of the Department of Human Genetics at Wits University, estimates the cumulative incidence by age 5 years of serious genetic disorders and birth defects at 5-8% of all annual births - 'nothing less than a silent epidemic'.

The 60,000 figure - or 5% of the 1.2 million babies born annually in South Africa - is a conservative estimate. Christianson says early recognisable genetic disorders and birth defects are the tip of the iceberg, because not all abnormalities like cystic fibrosis or congenital cardiac defects, are immediately detected.

By comparison, international studies indicate that 2-3% of all children are born with birth defects or genetic disorder recognisable at birth or shortly thereafter.

Christianson extrapolated the figures from a birth defect surveillance study in Limpopo province. Studies in other provinces where fetal alcohol syndrome is prevalent may have a significantly higher prevalence of genetic disorders and birth defects.

Serious defects may occur in 5-8% of births - 'nothing less than a silent epidemic'.

The Limpopo figures were worrying and the first steps have been taken to address the problem. In August 2001 the Health Minister released the government’s Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defect and Disability.

South Africa’s first locally trained and certified ‘sub-specialist’ in medical genetics, paediatrician Dr Louisa Bhengu, registered with the Health Professions Council in December 2002 and began working in poor communities in early 2003.

Professor Denis Viljoen, head of Human Genetics at Wits, said that ‘60,000 is the size of the burden, before we even touch other genetically determined disorders of later life such as diabetes, hypertension and cancer - we need another 30 Louisa’s and the funding to back them’.

The WHO requirement measure for medical geneticists is one per one million persons, which would put South Africa’s requirements at 40.

Viljoen says there are eight currently registered with the HPCSA, all of whom are working full-time, and five ‘part-timers who are retired or work in their primary disciplines like internal medicine and paediatrics but ‘do a bit of genetics on the side’.

Unlike Bhengu, all were registered on a grandfather clause after working in the field for years and were chosen by a committee of the SASociety for Human Genetics.

Bhengu will be employed as a community geneticist by the National Health Laboratory Services and make her academic and community contributions through Viljoen’s department.

‘Her relevance to underprivileged and underserviced communities through secondary hospitals and in primary health care around Gauteng will be immense. She’ll be helping to establish primary and secondary health care based genetic services for the care and prevention of genetic disorders and birth defects in communities that were previously disadvantaged and remain underserved,’ he said.

Viljoen said it was the first time that a training unit for medical genetics had trained and qualified a medical geneticist under the new rules of the HPCSA. The work crossed multiple disciplines and is the fastest growing biological discipline in science.

Bhengu, who was the liaising doctor between paediatricians and geneticists at Wits two years ago, was head-hunted. ‘We recognised her excellent potential and brought her into the department for training, which was hugely intensive. I encouraged as many people as possible in academic medical genetics in South Africa to participate in her training,’ said Viljoen.
An enthusiastic Bhengu is keen to begin a medical and public awareness campaign as an effective weapon in her armament against genetic disorders and birth defects. ‘Simple and cheap interventions for prevention are available, like folic acid supplementation that can reduce neural tube defect birth prevalence by up to 85%, as well as preventing other abnormalities including cleft lip and palate and heart defects. Primary preventive strategies are also available for common disorders including Down syndrome, fetal alcohol syndrome and congenital rubella.’

As part of a public awareness campaign, she also wants to target young women and grandmothers who care for children. Perhaps the best way of educating young women about these issues is to include coverage in the high school curriculum.

Bhengu expects to see the HIV pandemic influence her practice. ‘It’s in the literature but we haven’t really recognised it yet’.

Prophylactic intervention for HIV and counselling will become part of her work, for example when doing counselling for procedures like amniocentesis.

At the time of this interview, she was writing exams for her Masters in genetic counselling. ‘I think community work will open up new areas of research and intervention. We can wait for the Human Genome Project but that may take a while. In the meantime, we can get going and implement at community level the knowledge we already have’, she chuckled.

Bhengu said chromosomal defects accounted for 6% of all birth defects, single gene disorders for 7.5%, while 39% were multi-factorial disorders 20-30%, intrauterine environmental factors (teratogens) 5-10% and the rest were unknown.

In South Africa the common four genetic disorders and birth defects are Down syndrom, albinism, neural tube defects and fetal alcohol syndrome.

A major aspect of Bhengu’s clinical research focused on the use of videometric, three-dimensional camera techniques as a diagnostic tool for fetal alcohol syndrome. This is being developed in South Africa by the SA Foundation for Alcohol Related Research (FARR) in collaboration with Professor Vaughan and his group at UCT’s Biomedical Research Unit.

It enables two trained operators to measure and potentially diagnose up to 100 pre-screened six- to eight-year-olds in one morning versus the tedious former method of clinicians painstakingly measuring the child’s facial features.

The pictures are downloaded onto a pre-programmed laptop computer that helps the clinicians make a quick diagnosis. Wits, which is pioneering this technique, is believed to be the only centre currently using this method.

The university training course is currently taking applicants for the two-year study post, which is supported by the FARR that was founded by Viljoen.

Christianson revealed that when he first drew up his 5-8% cumulative incidence figures he was roundly criticised. ‘They all told me I couldn’t count and asked where these patients were. Well, it’s since been shown what we suspected - that the majority of them are dead. They’ve died without being diagnosed and thus recognised for what they represent - it’s simply just not put down on their death certificates that they had genetic disorders or birth defects’, he said.

He gave the example of two-thirds of all black children with Down syndrome in South Africa dying by the age of two.

The journey to turn this situation around and give medical genetics its rightful place in South African medicine will obviously be long, but is infinitely worth taking.

Chris Bateman

100 years ago: Partners in hospitals

At the February meeting of the Board of the New Somerset Hospital, the question was raised as to whether two partners in practice should be eligible to serve on the hospital staff at the same time. Now, of course, in the large hospitals at teaching centres in the United Kingdom the question cannot arise, inasmuch as the staff is drawn from a class of medical men who do not practise in partnership. In smaller hospitals, partly or wholly officered by GPs, it might come up, but so far as our experience goes, it is an unwritten rule well nigh universal, that partners should not serve together. In some cases we know it to be a rule formally embodied in the hospital bylaws. We remember one case some years ago in a small hospital, of a partner of a staff member being appointed with the result of a strong protest on the part of other members of the staff, which was supported by the local faculty. If we remember rightly, the appointment was cancelled.

(SAMJ March 1903, Pg 10)