

Down syndrome in paediatric outpatient wards at Durban hospitals

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To the Editor: The birth of a child with Down syndrome (DS) has many serious implications – physical and mental deficiencies, medical conditions and lifestyle challenges.¹ The high incidence (1 in 600 births in South Africa²) and high cost of medical care highlight the need for effective health care.² The South African Human Genetic Policy Guidelines include a rudimentary list pertaining to the management of DS.² In the USA, the American Academy of Pediatrics (AAP) has addressed this issue by devising clinical protocols³ focusing on improving the level of functioning and quality of life of children with DS. Countries such as Thailand⁴ and, to a lesser extent, South Africa, in KwaZulu-Natal,² have adopted and adapted these guidelines. The importance of the guidelines for clinical and counselling practices and their routine adherence is a daily challenge for 3 Durban hospitals with a total of 2 staff members dedicated to genetic matters.

DS studies^{4,5} have recognised the value of routine screening tests and genetic counselling, as they pre-empt the need for targeted therapy, early intervention programmes and support groups, which results in better quality of health and significant developmental progress.

We investigated and compared the clinical management of DS children at King Edward VIII, Prince Mshiyeni and Inkosi Albert Luthuli Central Hospitals in Durban with that occurring in countries such as Thailand, where the guidelines are implemented.⁴

Method

Medical chart checklists in accordance with AAP guidelines³ (Table I) were used to record information from hospital charts (Biomedical Research Ethics ref. no. BE100/08), to compare the care received by DS children and their families at Durban hospitals with the AAP recommendations and that observed in the Thai study.⁴ The 72 most recently treated cases of DS were assessed, and the relevant clinical staff facilitated the provision of further patient information including observation of routine patient practice in dealing with DS.

The data were captured on a Microsoft Excel spreadsheet, which allowed simple derivation of the frequencies of DS in maternal age groups, patient age groups, screening tests and counselling performed. The frequencies of screening tests conducted and genetic

counselling services offered were used as a measure of efficiency of health care and compared with AAP recommendations and findings reported by a study conducted in Thailand.⁴

Results

Table II shows the standard of clinical care offered to paediatric DS patients in this study in terms of screening tests and genetic counselling services, and a comparison with that observed in a Thai study⁴ and the AAP recommendations.³ The Thai study did not have recorded data for antenatal counselling and support group referrals.

Discussion

Irregularly performed screening tests and minimal genetic counselling indicate a need for specific DS guidelines for the strict adherence and employment criteria of genetic staff.

The SA guidelines² on the clinical management of DS offer a generalised list rather than a specific protocol. They include points addressing advanced maternal age, prenatal and postnatal diagnosis, preventive measures, rehabilitation and genetic counselling. Although relevant, there are no specific and utilisable clinical protocols on the management of DS patients in terms of the types of screening tests, preventive measures and rehabilitation. They also lack focus on the factors of age groups and development of DS patients.

Our study indicated a significant difference between AAP guidelines (Table I), the statistics from the Thai study, and the observed frequencies of tests and services offered cumulatively at the three Durban hospitals (Table II). While the Thai study adhered to the AAP recommendations, the Durban hospitals lagged in both screening tests (thyroid function, hearing and vision) and genetic counselling services (antenatal and support groups). However, screening tests for cardiac defects were performed often (84.7%) owing to their frequent occurrence and cause of morbidity and mortality in DS.⁶

Another concern is the large number of mothers (42.9%) of advanced maternal age and low frequency of antenatal counselling

Table I. AAP guidelines for management of DS children at developmentally significant age groups

Patient age group	Clinical focus
Prenatal	Genetic counselling on whether to continue or terminate pregnancy
Newborn (birth - 1 month)	Clinical manifestations, developmental progress (eye function, feeding, hearing ability), support groups, coping methods
Infancy (1 month - 1 year)	Screening tests conducted for hearing, vision and thyroid function and annually thereafter. Growth and development is monitored
Early childhood (1 - 5 years)	Physical, occupational and speech therapy. Diet and exercise regimen is monitored
Late childhood (5 - 13 years)	Physical and sexual development

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Table II. Comparison of frequencies of screening tests and genetic counselling services offered in this study with AAP recommendations and a Thai study

	Recommended (%)	Observed in Thai study (%)	Observed in present study (%)
Screening tests			
Hearing	100	95	56
Vision	100	94	56
Thyroid function	100	100	18.1
Cardiac defects	100	100	84.7
Genetic counselling services			
Antenatal counselling	100		11.1
Postnatal counselling	100	100	84.7
Referral to support group	100		27.8

p<0.5; statistically not significant.

observed in the study. Advanced maternal age (a crucial factor in DS, illustrated by 80% of births being to mothers >35 years old) should be considered in a resource-poor setting such as South Africa.

Regarding genetic staff, there was during the study one full-time geneticist, one part-time geneticist, and no genetic counsellors employed across the three hospitals – a stark contrast to the recommendations made by the Human Genetics Policy of SA which prescribes a minimum of 36 genetic counsellors and 18 medical geneticists for KZN.² The subsequent inadequate clinical care of DS patients is highlighted in this study and has prompted media attention and lawsuits for negligence.⁷

The provincial and national health departments bear the ultimate responsibility for implementing an agreed set of protocols that are strictly adhered to, and for employing sufficient genetic staff to ensure effective health care for DS patients and their families. Besides the financial benefit of a largely preventive health care programme, the quality of life of DS patients and their families will increase exponentially while alleviating the strain on the health care system.

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