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Review

Anorectal malformations in Africa

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Anorectal malformations (ARMs) remain a significant birth defect with an accepted incidence of approximately 0.2 - 1.2%.[1,2] Although major advances have occurred in the management of these children during the last 15 years, ARMs remain a clinical challenge largely because of the significant reconstructive and management aspects involved, many of which are related to faecal and urinary continence and possible sexual inadequacy in later life.

Geographical variation occurs in the overall incidence, individual phenotypes and regional geographical subtypes of ARM. In Europe, inter-registry incidence may vary from 1.14 to 5.96 per 10 000,[3] with geographical yearly fluctuations reported.[4] Although variations may be based on environmental and socio-economic factors, genetics may have a significant role to play. Ethnic differences therefore appear to exist both in incidence and type of ARM.[5-7] It is possible that there is a lower incidence of congenital malformations and syndromes as well as a higher incidence of low lesions in populations in developing countries.

Early North American studies indicated a low incidence of anal atresia in black patients.[5,8] It is not clear whether this is specific to North America or whether it is generally true, as other studies have shown no clear-cut distinction in incidence in black patients.[9] In developing countries, black patients frequently live in rural communities where health needs are under-resourced, and a measure of underreporting could be present.

Despite early suggestions to the contrary, there is a body of evidence suggesting that ARM is not only a significant clinical load in Africa[10,11] but may in fact be more common in the African population.[12,13] There is as yet inadequate objective evidence to support this hypothesis and also a great paucity of knowledge as to the types, frequency and incidence of ARMs and their associated anomalies encountered on the African continent.

Louw[10,12] reported an incidence of 1:1 740 among whites, 1:1 770 among coloureds and 1:2 260 among blacks in Cape Town, which is higher than the 1:5 000 incidence reported elsewhere.[5] Shija[11] reported on 46 patients seen during a 2-year 'sabbatical' in Zimbabwe, suggesting that ARM was fairly common in that region. This has since been borne out by other workers in Africa, and personal communications from Tanzania, Zimbabwe, Kenya, Uganda, Ghana and Nigeria indicate that ARM constitutes a significant surgical load in Africa. In Nigeria ARM has been identified as the third most common cause of neonatal intestinal obstruction, representing 13.4 - 20% of congenital malformations.[14,15] ARM also represented 67% of neonatal emergency surgical procedures.[16]

In addition to variations in overall incidence, ethnicity and genetic influences may result in individual variations, associated anomalies and different patterns of disease. In our current South African study, preliminary data on more than 1 000 ARM patients show a relatively high prevalence in all ethnic groups including black patients. Although ARM appears to be less frequent in black patients in our unit in the Western Cape (approximately 20%) (S W Moore *et al.* – unpublished data), a very large series from another centre (KwaZulu-Natal) consisted of 86% black patients. It would therefore appear to be mainly an expression of demographics.

The 1.02 male-to-female ratio of black patients in this study is similar to the overall incidence (1.1) and is similar to ratios in other ARM studies.[3,17,18] Exceptions include anal atresia (male preponderance) and ectopic anus and congenital anal and other fistulas (female preponderance).[3,19]

Forty-one per cent of lesions in the black group were considered high (supralevator). Eighty per cent of black females had low lesions; 210 (74%) were lesions of the vestibular anus, and 24 (8.4%) were perineal fistulas. Thirty-five black patients with high lesions had cloaca (3.8%). This is in contrast to a report of 44.4% low (or anal) lesions reported from Nigeria[20] and approximates the reported world experience.[17] This illustrates the need for more objective data from developing countries to assess geographical differences. A multicentre study is currently underway to study the epidemiological prevalence of ARM in Africa.

Apart from a much lower incidence of chromosomal lesions (1.5% v. an 8 - 15% incidence) and an apparent increase in cloacal abnormalities (3.8%) the incidence of associated anomalies was more or less equal to international norms. Black patients had a 38% overall prevalence rate of associated anomalies. The most common anomalies encountered in black patients were genito-urinary (26%), and included renal agenesis, multicystic kidney and hydronephrosis, and vaginal anomalies (e.g. absent or double vagina). The VACTERL (vertebral, anorectal, cardiac, tracheo-oesophageal, renal and limb abnormalities) group of anomalies were next most common including vertebral anomalies (14%), oesophageal atresia (13%), cardiac anomalies (13%) and skeletal anomalies (10%), but only 6 had a full VACTERL association. A fairly constant association was noted with gastro-intestinal malformations (10%) which included malrotation (3%) and Hirschsprung's disease (0.4%). Other

anomalies included those of the external genitalia, notably hypospadias (9%), anterior abdominal wall defects (5%), facial anomalies (3%) and 11 patients with dysmorphic features (Down syndrome 2). Neuroblastoma was associated in 1 patient.

The definition and classification of ARM remains a historical problem. In his study Smith[13] reported that 47% of patients had rectal anomalies and 53% anal anomalies. In an attempt to set standards for comparison, the Wingspread classification in 1984[13] and a recent international consultation[21] adopted a fairly robust simple classification that should be achievable by most developing countries (Table I). All efforts should be made to achieve uniformity so as to be able to compare similar groups across geographical and ethnic boundaries.

TABLE I. NEW PROPOSED INTERNATIONAL (KRICKENBECK) CLASSIFICATION OF ANORECTAL MALFORMATIONS[21]

Frequent anomalies	Rare/ regional variants
Perineal (cutaneous) fistula	**'Pouch colon'**
Recto-urethral fistulas	**Rectal atresia/stenosis**
Bulbo-urethral	**Rectovaginal fistula**
Prostatic	**H-fistula**
Rectovesical fistula	**Others**
Vestibular fistula	
Cloacae	
No fistula	
Anal stenosis	

The search for the aetiology of ARM remains an enigma. Although environmental factors may play a role, genetics has emerged as a strong contributing factor despite the relatively low familial incidence.[13] Candidate genes have been reported in at least two rare syndromes, viz. Currarino[22] and VACTERL associations.[23] ARM-related syndromes often represent developmental 'field defects'.[24] The role of fundamental signalling pathways and endothelin involvement have been reported.[25] This is of considerable interest with regard to black patients as parallel functions have been demonstrated in melanoblast development.[26]

In summary, according to our current state of knowledge ARM does not appear to be uncommon in black populations in Africa but constitutes a significant clinical and surgical load. Further study is required to look at a possible increased frequency in black African populations. Local variations in subtype distribution may occur and associated anomalies are not uncommon. There is as yet insufficient evidence to suggest a variation from the international norm. Disrupted genetically related signalling pathways appear to be the most likely aetiological factor and should be further investigated in African populations.

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