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Congenital Adrenal Hyperplasia in Sudanese Children

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Objective

To collect baseline data on congenital adrenal hyperplasia (CAH) in children in Sudan.

Design

A retrospective study conducted in two major referral hospitals in Sudan. Medical records of all confirmed cases of CAH over a 9.5 year period were reviewed. A semi-structured data collection sheet containing demographic, clinical, and social information was used.

Results

101 patients with CAH were confirmed, of which, there were 68 cases of salt-wasting 21-hydroxylase deficiency (21-OHD), 21 cases of simple virilising 21-OHD, 9 cases of 11-beta hydroxlase deficiency, and 3 cases of 3-beta hydroxy steroid dehydrogenase deficiency. 75% of cases were genotypically females (46,XX) with a female to male ratio of 3:1, meaning that around 50 males must have died with unrecognized CAH during the study period. Parental consanguinity was reported in 88.5% of cases. Unexplained infant/neonatal deaths in siblings of patients were found in 36.5% of cases. As per the initial gender assignment, the wrong gender was assigned initially to the baby or child in 31% of cases with the subsequent need for reassignment. Overall the mean duration between onset of symptoms and diagnosis was 17.3 months. Out of the cases having ambiguous genitalia at birth or as first presentation (68%), 45% were wrongly reassured by either birth-attending midwives or doctors instead of being referred to specialized care.

Conclusion

Urgent measures to spread the awareness of the condition among child-health care providers are needed to prevent male deaths and reduce the need for sex reassignment in females with CAH in Sudan.