Editorial Comment

Bridging knowledge gap on birth defects

Birth defects, reported to affect an estimated 1 in 33 infants, are common but documentation of the size of the problem, as with many other health conditions in sub-Saharan Africa, is rather limited. Having a baby with congenital malformation, however mild, is psychologically disturbing and a source of concern to parents and other family members. Apart from high case fatality as in the case of anencephaly, the endless visits to medical practitioners for any intervention have a toll on the quality of life of families in general. In this issue of the journal, two manuscripts highlight the peculiar problems with diagnosing babies with birth defects. Using ultrasound screening, a multidisciplinary team of researchers comprising Paediatricians, various surgical specialists, Obstetricians and Gynaecologists and Radiologists documented anomalies in 7.8% of the cohort studied which is more than two times the usually reported prevalence of the disorder. The frequencies of congenital anomalies in various systems were documented, albeit from a retrospective angle.

It is important as a follow up that mechanistic studies should take place for determining the "why" and "how" of the disorder. Consanguinity was excluded by the authors as this is not a common practice in the region for cultural reasons. However, in this era of genomics, other genetic factors should be explored. Thus collaboration between clinical and basic scientists should always be advocated for solving the puzzle of birth defects. The authors working together highlight the point that the era of researchers working "in silos" is over and a team spirit should be engendered. That is the only was various facets of a medical problem can be dealt with and a 360 degree approach achieved. For developing countries to make the right impact on health, multidisciplinary teams should collaborate in tackling the challenges.

The very low prenatal diagnosis of less than 1% implies that better screening programmes of pregnant mothers are needed. The use of amniocentesis and biomarker assays in at-risks individuals as well as improvement in other diagnostic facilities should be the standard practice. Ultrasonography combined with magnetic resonance imaging of higher resolution than presently available will improve diagnostic specificity. This was convincingly discussed by the team of researchers. We need more disease registries on various health conditions and congenital anomalies are important enough to be included on such a list. This is useful for international data comparison on epidemiology, observing time trends, disease outcomes and for health systems research. This is an essential take-home message from the authors.

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