Original Article

Consanguinity and Disorders of Sexual Developments in the Sudan

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Abstract:

Background: Consanguinity is very common in the Sudanese society. There is a lack of studies on consanguinity and its impact on genetic diseases in Sudan. In this study we correlated Disorders of Sexual developments (DSDs), as an example of genetic conditions, in relation with consanguinity.

Material and Methods: A cohort of twenty six cases from 15 Sudanese families were diagnosed with DSD between the years 2008-2010, Diagnosis was done in Al-Neelian Medical Research Centre, Al-Neelain University, Sudan in collaboration with the University of Lübeck, Germany.

Results: Parental consanguinity was seen in 70 %, 10 % were not consanguineous while 20 % did not provide a detailed family pedigree.

Conclusion: There is strong association between consanguinity and inheritance of DSDs in the Sudan, which is expected to be higher than that reported in the literature about non-consanguineous DSDs from western countries.

Keywords: genetic, chromosomal, congenital.

Consanguineous marriages are frequent in many countries due to cultural perspectives, with first cousin marriage being the most common[1]. Available data suggest that genetic and congenital disorders are more common in Arab countries than in industrialized countries and recessively inherited disorders are the main cause of physical and mental handicap[2-6]. Congenital malformations occur more commonly in the Middle East region than in Western countries and are responsible for high morbidity and mortality rates[7-10]. Studies on the relationship between consanguinity and genetic disorders have been reported from many parts of the world but there are very few publications that come from Arabic and African countries.

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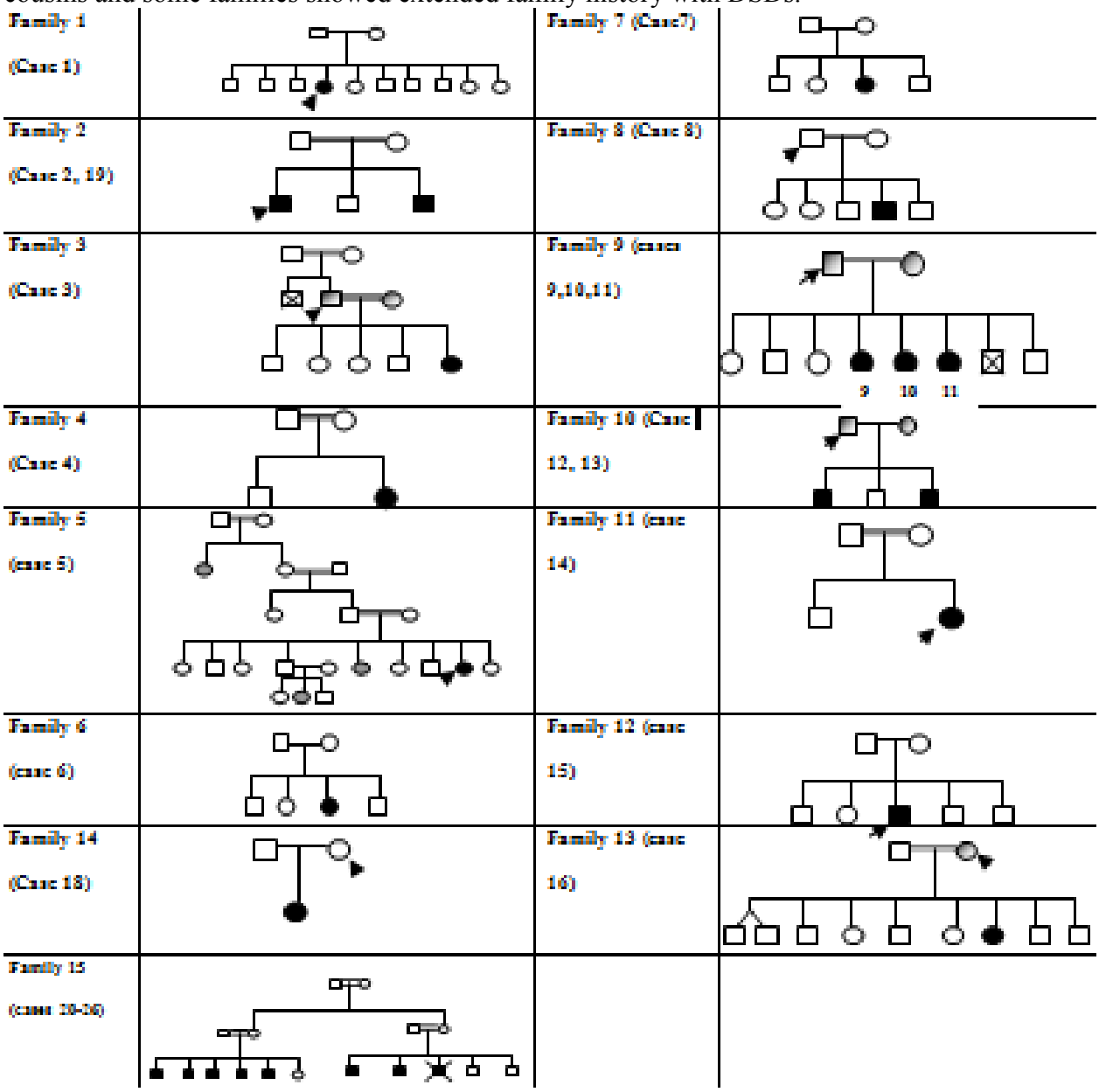
Common examples for heritable genetic disorders are Disorders of Sexual Developments (DSDs). DSDs are defined as a congenital condition in which development of chromosomal, gonadal, or anatomical sex is atypical[11]. Patients usually present with ambiguous external genitalia. The incidence of the disorder is 1:4,500 to 1:5,000 live births[12, 13]. In Egypt they estimate the prevalence to higher and approach 1: 3,000 live-births[14]. Depending on the underlying disorder, they may either follow X-chromosomal recessive, or autosomal-dominant, or autosomal-recessive inheritance. Autosomal-recessive inheritance is expected to be high in societies with consanguinity marriages among carriers of the related mutation. Therefore, we would expect the spectrum of the different forms of DSDs to be different in Sudan from other countries, especially studies from Western societies reported in the literature. There are no records about the prevalence of DSDs in other Arab or African countries. In this study we explored the prevalence of consanguinity in the Sudan on the occurrence of Disorders of Sexual Developments as an example of genetic disease.

Table 1: Pedigrees from the participating families in this study as most of the parents are first class cousins and some families showed extended family history with DSDs.



□	Un affected male	○	Un affected male
◊	Affected female	▪	Affected male
▫	heterozygous	⬭	heterozygous
■	Homozygous	●	Homozygous
⊠	Dead	⊗	dead

Materials and Methods:

During two years (2008-2010), twenty six patients were referred to Al-Neelain Medical Research Centre, Faculty of Medicine and Health Sciences, Al-Neelain University.

Genetic analysis confirmed all patients have one of the different types of DSDs, inherited in an autsosomal recessive pattern. Family history was investigated and parental

consanguinity among the different referred cases was drawn on a family tree (Table 1).

Results: The parents of family 1 and 15, 14 were not consanguineous. The parents from family 4 and 6 were first class cousins, while in families 2, 3, 5, 7, 8, 9, 10, 13 they were second class cousins and in family 15 they were third class cousins. Unfortunately case 17 did provide neither family history nor information about parental consanguinity.

Discussion:

The role of consanguinity as a cause for congenital malformations has been studied by several researchers from countries with consanguinity rate[7, 9, 15-17]. Consanguineous marriages have a comparatively higher risk of producing offspring with genetic damage than that of the general population. Accordingly, the occurrence of autosomal-recessive genetic diseases is expected to be higher in consanguineous marriages. In countries such as Sudan, with a high consanguinity rate, it is also alluring to blame consanguinity as one of the causes of conditions with a genetic basis without the need of evidence of causation. This study is clarify the role of consanguinity as a causative factor in the incidence of DSDs. The likelihood of detecting an effect of consanguinity among the first class cousins were high as well as among cases within second and third class cousins as well. This is because usually they are related in their back-ground family which makes the genetic relation stronger. Proving that genetic disease such as DSDs can also occur in second and third class cousins as well. DSD can also occur sporadically and idiopathic in a non-consanguineous marriages, although not as common as in consanguineous marriages. Therefore, the differential diagnosis of DSD in Sudan will be different from the Western countries, and diagnostic analysis should be adjusted accordingly.

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