ABSTRACT

In The Medical Genetic Unit of King Abdulaziz University in Jeddah was established by the author in February 2005. It contains the first genetic clinics in the Western region of the Kingdom of Saudi Arabia. Jeddah, the commercial capital and the second largest city of Saudi Arabia, has a population of over 3.5 million people with a 2.06% growth rate and the median age of 21.4 years. Referrals are received from Jeddah and surrounding cities and towns in the Western Region including among others Makkah, Taif, and Al-Baha. The objectives of this prospective study were to study the pattern of genetic disorders over a 4 year period, elucidate their inheritance patterns, set up a platform for a genetic database in the region and identify the relevant risk factors for genetic conditions in our population, especially those which are potentially preventable. Nine hundred and four patients with genetic disease have been assessed. Each patient received a full genetic assessment including a dysmorphological examination. Imaging and laboratory testing (including genetic testing) were done as appropriate. The most likely inheritance pattern was based on the diagnosis and analysis of family pedigree. The most common form of genetic disease was autosomal recessive followed by chromosomal disorders and then autosomal dominant disorders. Unlike some other reports from Saudi Arabia, consanguinity was more likely to be present in individuals with autosomal recessive disorders than with any other form of genetic disease including isolated congenital malformations. Recommendations to decrease the burden of genetic disease in this region including our experience with the current Premarital Screening in our student population are discussed.

BACKGROUND

Population of Saudi Arabia is a mixture of ethnic groups
Specific genetics and cultural factors
Consanguinity rates: 2560%
Diminished awareness of the importance of genetic counseling in preventing malformations

JEDDAH

Cosmopolitan city
Population >3 million
Commercial capital of Saudi Arabia
Western coast of the Red Sea
Multicultural society
Heterogeneous population

OBJECTIVES

Study the pattern of genetic disorders at the western region of KSA over a 24 month period
Elucidate the inheritance patterns for these genetic disorders
Determine the relevant risk factors for genetic conditions in this population- especially those that are potentially preventable
Provide recommendations for reduction of genetic disorders- role of genetic counseling and feasibility of antenatal diagnosis

CONCLUSIONS:

- Estimated recurrence risk of genetic disorders is > 1% in almost all cases and more than half RR ≥ 25%.
- Overall 32.61% of all cases had a RR >1% AND Antenatal diagnosis was possible and feasible
- The need for genetic counseling is underscored by the fact that 55.4% of mothers were <36 years and likely to have further children.
- Factors amenable to premarital and preconception counseling and more common in the region are consanguinity and advanced maternal age.
- A condensed public education program in addition to education of all health care providers who can play an essential role will be highly cost effective in reducing the burden in genetic disorders in the KSA.