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RESEARCH ARTICLE

THE MOST COMMON GENETIC DISEASE IN HAIL AND DISEASES RELATED TO CONSANGUINITY.

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Abstract

Background and objective: There are common genetic diseases in Hail,but the information about it are still limited.The objective of this cross-sectional study was to detect the most common genetic disease and the risk factor of consanguinity on genetic diseases in Hail.

Subjects and methods: The study samples were determined by multistage probability random sampling procedure. Genetic diseases were obtained from 500 papers and electronic questionnaires. The data were analyzed by using SPSS and calculator.

Results: The proportions of genetic diseases were ordered from highest to lowest as follow: Type 1 Diabetes Mellitus(20%), Congenital Heart Disease(9%), breast cancer(8.2%), Down syndrome (6.4%),Sickle cell disease(3.8%), Muscular Dystrophy (1.8%) and Thalassemia (1%).However, 49.6% of sample answered the question were consanguineous. The proportions showed Thalassemia was the highest disease affected by consanguinity relationship with (100%, *p= 0.023*),followed Sickle cell disease (73.68%,*p= 0.032*) andbreast cancer (34.14%,*p= 0.039*).

Conclusion: The data suggested that the most common genetic disease in Hail is Type1 Diabetes Mellitus and consanguinity is one of the risk factor on some of these genetic diseases as Thalassemia, Sickle cell disease and breast cancer.

Introduction:-

Quick search for published genetic diagnoses from Saudi Arabia readily reveals the clear bias toward autosomal recessive disorders [1] such as, Sickle cell Disease (SCD) and Thalassemia. The spread of these genetic blood disorders differs among the various regions of the Kingdom. According to the statistics released by the Ministry of Health (Health Marriage Against Genetic Blood Disorders Program: SCD and Thalassemia), from 1425H till the end of 1430H, the recorded incidence of SCD was 0.27%, whereas the incidence of thalassemia was 0.05%.

However, the Saudi Premarital Screening Program estimated the prevalence of the sickle cell gene in the adult population at 4.2% for sickle-cell trait and 0.26% for SCD, with the highest prevalence noted in the Eastern province (approximately 17% for sickle-cell trait and 1.2% for SCD) during February 2004 to January 2005[2], but in the children and adolescentsSCDwas detected in 108 of 45,682 with a prevalence of 24 per 10,000. The regional distribution of SCDshowed eastern region dominance with a prevalence of 145 per 10,000, followed by the southern

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region with a prevalence of 24 per 10,000, western region 12 per 10,000, and central region with 6 per 10,000. No cases were found in the northern regions. The male to female ratio was approximately 1:1 during 2004 to 2005 [3]. Prevalence of beta thalassemia trait are 3.22% and thalassemia disease had 0.07% mainly in the eastern, western, and southwestern parts of the country,and it was taken from 488,315 individuals screened in February 2004 to January 2005 [2].

Other genetic diseases as Down syndrome which is one of the most common chromosomal disorders in Saudi Arabia, its prevalence was 1.8 per 1000 since, July 1982 to June 1991[4]. Congenital Heart Disease (CHD)in Saudi Arabia has been reported in 4 regions of Saudi Arabia in (August 1988-February 2000) and 2604 individuals with CHD were evaluated. Ventricular septal defect was the commonest lesion (33.9%) followed by atrial septal defect (18.1%) and the sex distribution was similar; for 3 conditions, more males than females were affected andof 2269 (59%) presenting in the first year of life, 566 (24.9%) had neonatal CHD [5].

Type 1 diabetes mellitus prevalence in Saudi Arabian in children and adolescents is 109.5 per 100,000 in 2001-2007. The male to female ratio was almost equal (26 males and 24 females) and its distribution by region shows that the highest was 162 in the central region, and the lowest was 48 in the eastern region. Children and adolescents were also grouped by age into 5-6 (prevalence 100), 7-12 (prevalence 109), 13-16 (prevalence 243), and 17-18 (prevalence 150) [6].

Muscular dystrophy which is X-linked genetic neuromuscular disease of childhood, it is relatively frequent, with an incidence is 1 in 3500 male live birth [7].

The yearly percentage distribution of incidence of malignant breast cancer showed a steady rise over the years, as compared to benign breast lesions. In 2000, 76.5% of the tissues submitted for histopathologywere benign and only 23.5% were malignant. From 2000 to 2007, there was a steady rise by a mean of 4.8% in the annual incidence of malignant breast lesions, with the exception of 2003, when there was a slight decrease. The annual rate of a malignant breast lesions confirmed by histopathological examination ranged from 23.5% in 2000 to 47.2% in 2007. In contrast, after 2007, there was a shift of the trend toward more benign cases. There was increase in the percentage of people diagnosed with breast cancer from 23.5% in 2000 to 34.5% in 2010 [8]. The Saudi Cancer Registry reported a rising proportion of Breast Cancer among women of all ages, from 10.2% in 2000 to 24.3% in 2005[9].

The high rate of consanguinity has greatly impacted the landscape of genetic disorders in Saudi Arabia, so the most common genetic disorders are SCD**,** Thalassemia, Down syndrome, CHD, type1 diabetes mellitus, muscular dystrophy and breast cancer [4].
In this recent study, we present the incidence of the most common genetic diseases in Hail region together with recommendations for developing strategies for prevention and health care system.

Methodology:-

In this study participants were selected by multistage random probability sampling of Hail households from each region. This cross sectional sample was used to know the most common genetic diseases in Hail. A questionnaire was designed for this purpose and administered to the families through paper questionnaires or electronic questionnaires, male (34) and female (466) with total number of (500). One member of each family was asked about personal data as age, sex and the consanguineous marriage for member and member parents. Also this questionnaire asked about cases of genetic diseases in these families, including inherited blood diseases as thalassemia and sickle cell anemia, chromosomal disorders as Down syndrome, multifactorial disorders as type 1 diabetes mellitus and breast cancer, and also muscular dystrophy and congenital anomalies as CHD.

Data were analyzed by using the SPSS software package, and the chi-square test was used to compare proportions of cases in consanguineous. A statistically significant difference was assumed when the $p < 0.05$.

Results:-

The highest proportion was 20% in Type 1 Diabetes Mellitus, accounting for (100) cases of genetic disorders from all collected samples. CHDwas the second high proportion, occurring in 9% of cases, whereas Breast Cancer occurs in 8.2%. Down syndrome included in 6.4% of samples, followed by SCD occurs in 3.8%. Muscular Dystrophy was

1.8%. Finally, the lowest proportion in this questionnaire was in Thalassemia with percentage of 1% as shown in Table 1.

In our study, 500 of sample answered the question on consanguinity (49.6%) were consanguineous. Table 2 shows pattern of genetic disorders and parental consanguinity, indicating that Thalassemia was the commonest type accounting for 100% of cases, because they were 5 and all those were consanguineous. Then, SCD has 73.68%. WhereasMuscular Dystrophy included55.55%of consanguinity relationship and diabetes mellitus Type 1 was 52%. Down Syndrome has 50% and CHD occupied 40% of cases. Finally, Breast Cancer was the least percent of consanguinity (34.14%).

Table 1:- Genetic disease in Hail city

Condition	Number of Condition	Percent %
Type 1 diabetes mellitus	**100**	**20%**
Congenital heart disease	**45**	**9%**
Breast Cancer	**41**	**8.2%**
Down syndrome	**32**	**6.4%**
Sickle cell disease	**19**	**3.8%**
Muscular dystrophy	**9**	**1.8%**
Thalassemia	**5**	**1%**

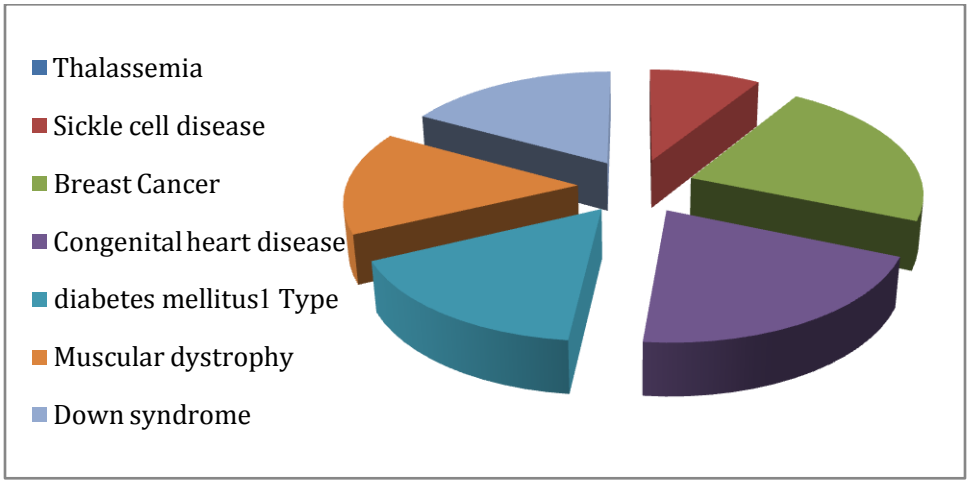

Fig 1:- the % distribution of most common genetic disease in Hail city

Table 2:- The relation of genetic disease and parental consanguinity in Hail city

Condition	Consanguinity	Non- Consanguinity	*P* value
Thalassemia	**5 (100%)**	**0 (0%)**	**0.023**
Sickle cell disease	**14 (73.68%)**	**5 (26.31%)**	**0.032**
Breast Cancer	**14 (34.14%)**	**27 (65.85%)**	**0.039**
Congenital heart disease	**18 (40%)**	**27 (60%)**	**0.178**
Type 1 diabetes mellitus	**52 (52%)**	**48 (48%)**	**0.592**
Muscular dystrophy	**5 (55.55%)**	**4 (44.44%)**	**0.719**
Down syndrome	**16 (50%)**	**16 (50%)**	**0.963**

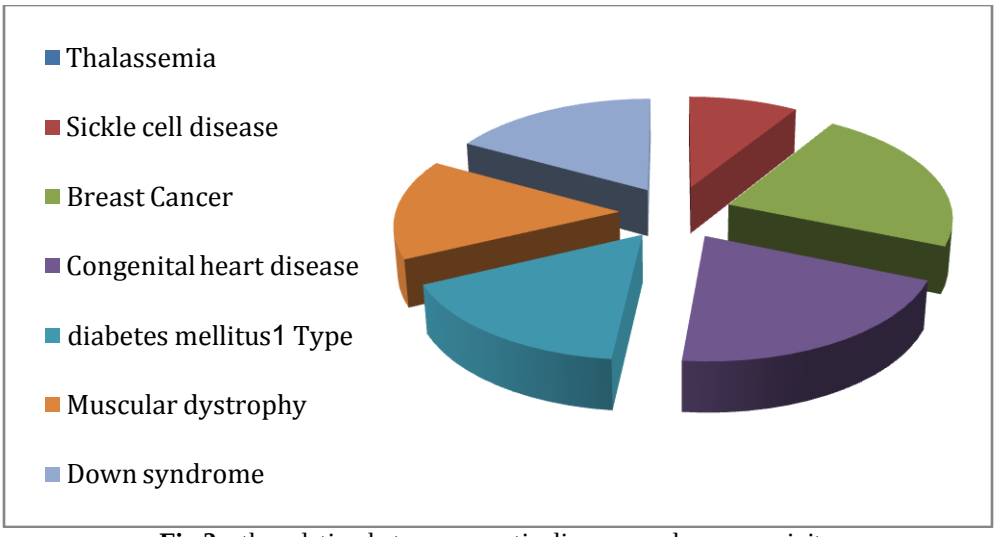

Fig 2:- the relation between genetic diseases and consanguinity

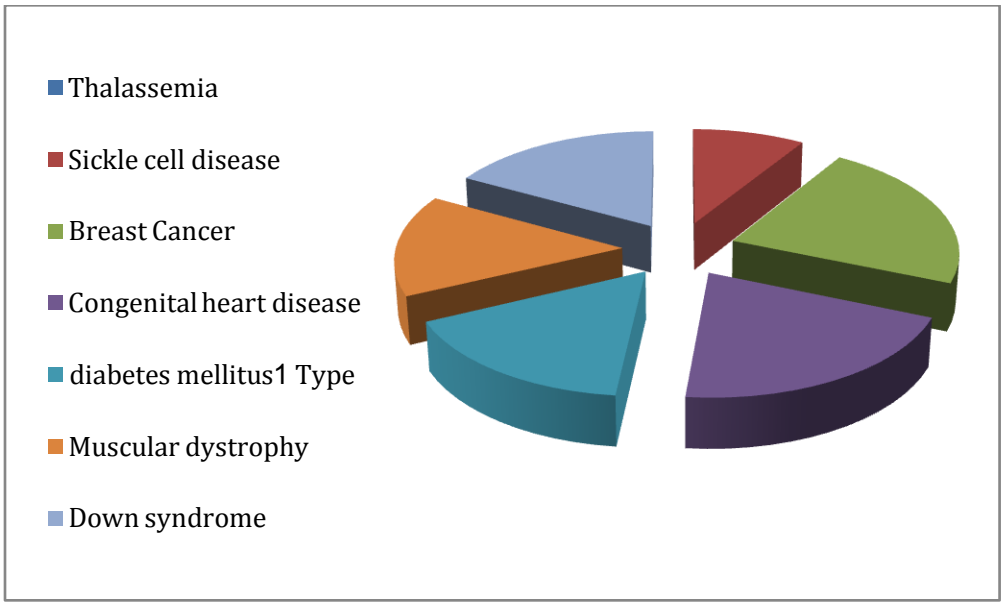

Fig 3:- the relation between genetic diseases and non-consanguinity

Discussion:-

When comparing the result of genetic disorders in this study to previously published studies, the demonstrated rates seems to be much higher in some diseases and lower in other diseases than studies done before. For examples, prevalence of SCD washighest in the Eastern region with 13.41% per 1000, followed by Southern and Western regions with 5.56% and 2.85%, respectively and lowest in Central and Northern regions with 1.37% and 1.35%, respectively[10].In more, the regional distribution of SCD showed Eastern region dominance with a prevalence of 1.45%, followed by the Southern region with a prevalence of 0.24%, Western region 0.12%, and central region with 0.06%. No cases were found in the Northern regions [3]. However, the proportion of SCD in Hail was lower than Eastern and Southern regions,but higher thanWestern Central and Northern regions.Thalassemia was highest in the Eastern region (5.9%), moderate in the Southern, Western and Central regions (1.42%, 1.02%, and 1.01% per 1000, respectively) and lowest in the Northern region with 0.39% [2]. In this study, Thalassemia was lower than Eastern, Southern, Western and Central regions,but higher than northern region.The registry of King Faisal Specialist Hospital and Research Centre reported that the number of Breast Cancer cases has increased considerably,there were 1152 female Breast Cancer cases in 2008 in comparison with 1308 in 2009, and 1473 in 2010. Breast cancer ranked first among females accounting for 27.4% of all newly diagnosed female cancers (5378) in the year

2010[8].Compared to the rate reported in the literature of Breast Cancer,we found Breast Cancer decreased than previous cases reported [9]. Our findings of Down's syndrome seem to be much higher than previous study[4]. Down's syndrome was ascertained in 0.18% of 23,261 consecutive babies born alive to Saudi women, giving an incidence of 1 in 554 live births (1.8 per 1,000)[4],so that lower than our findings of (6.4%). Muscular dystrophy, it is relatively high than study done before[7].In Alqassim region,the pediatric cardiology unit of King Fahad Specialist,Buraidah had 320 of 379 patients with CHD,so the CHD in Alqassim lower than in Hailwhich Hail has 45 CHD of 500 samples [11]. Type 1 diabetes mellitus was not related with consanguinity [12] and this agree about type 1 diabetes mellitus with our study.

Theoretically, consanguineous marriages have a relatively higher risk of producing offspring with genetic damage than that of general population. Accordingly, the concurrence of genetic disease should be higher in consanguineous marriages[4]. In countries,such as Saudi Arabia with a high consanguinity rate,it is tempting to blame consanguinity as one of the causes of condition with genetic diseases, so in this study SCD, Thalassemia, Down syndrome and type1 diabetes mellitus were high in consanguineous marriages.

Recommendation:-

Premarital screening to detect the Hb S (SCD) or Thalassemia to both partners. If they are carriers, they must have Genetic counseling trying the birth prevention of an affected child .Screening for presymptomatic individuals who are at risk for diabetes and breast cancer. Doing the triple test for the detection of Down Syndrome in the pregnant woman.

References:-

1. Alkuraya FS. Genetics and genomic medicine in Saudi Arabia.*Mol Genet &GenomicMed2014;2(5): 369–378.*
2. AlHamdan NA, AlMazrou YY, AlSwaidi FM and Choudhry AJ. Premarital screening for thalassemia and sickle cell disease in Saudi Arabia. *Genet Med 2007; 9:372–377.*
3. Al-Qurashi MM, El-Mouzan MI, Al-Herbish AS, Al-Salloum AA and Al-Omar AA. The prevalence of sickle cell disease in Saudi children and adolescents. A community-based survey. *Saudi Med J2008; 29:1480–1483.*
4. Niazi MA1, al-Mazyad AS, al-Husain MA, al-Mofada SM, al-Zamil FA, Khashoggi TY and al-Eissa YA. Down's syndrome in Saudi Arabia: incidence and cytogenetics. *Hum Hered 1995; 45(2):65-69.*
5. Alabdulgader AA1.Congenital heart disease in Saudi Arabia: current epidemiology and future projections.*EastMediterr Health J 2006; 12 Suppl 2:S157-167.*
6. Al-Herbish AS1, El-Mouzan MI, Al-Salloum AA, Al-Qurachi MM and Al-Omar AA.Prevalence of type 1 diabetes mellitus in Saudi Arabian children and adolescents. *Saudi Med J 2008; 29(9):1285-1288.*
7. Tayeb MT. Deletion mutations in Duchenne muscular dystrophy (DMD) in Western Saudi children. *Saudi J Biol Sci 2010; 17(3): 237–240.*
8. Al-Rikabi A and Husain S. Increasing prevalence of breast cancer among Saudi patients attending a tertiary referral hospital: a retrospective epidemiologic study. *Croat Med J 2012; 53(3): 239–243.*
9. Dastgiri S. Frequency, incidence, and prevalence. *Saudi Med J 2016; 37(3): 329.*
10. Memish ZA, Owaidah T M andSaeedi M Y. Marked regional variations in the prevalence of sickle cell disease and β-Thalassemia in Saudi Arabia.*J Epidemiol and Glob Heal2011; 1(1): 61-68.*
11. Jaiyesimi F , Ruberu DK and Misra VK. Pattern of congenital heart disease in King Fahd Specialist Hospital, Buraidah.*Ann Saudi Med1993; 13(5):407-411.*
12. El Mouzan MI,Al SalloumAA, Al HerbishAS, Qurachi MM andAl Omar AA. Consanguinity and major genetic disorders in Saudi children: a community-based cross- sectional study. *Ann Saudi Med 2008; 28(3): 169-173.*