Characteristic Factors Affecting Utilization Patterns of Prenatal Genetic Services in the Bedouin Population

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Background

The practice of marriage between close relatives increases the risk of genetic disease. The Bedouin population in southern Israel is a tribal, traditional, Arab-Muslim cohort that is characterized by high consanguinity. Because of that, autosomal recessive genetic diseases are prevalent in this community, causing high infant mortality rates. Over thirty genetic diseases are known among this population, and many of them can be identified through surveys and diagnostic carrier tests.

The Bedouin population is offered the genetic screening tests free as part of the Israeli public health services. Whereas until 2010 genetic screening tests were offered to only some of the tribes in the Negev, beginning November 2010, all women of childbearing age were offered carrier screening for hypothyroidism – retardation – dysmorphism syndrome (HRD), as well as for tribe-specific diseases. In recent years there have been efforts to increase access to genetics prevention programs and adapt them to the population.

The main change is the introduction of a community nursing genetic services (CNGS) in the health care system, targeting all couples of reproductive age, to effectively disseminate information on genetic screening tests, and facilitate implementation of testing for those who choose to do so after counseling. In this study we examine the efficiency and accessibility of the community genetic services, and their impact on uptake of genetic carrier screening in community health clinics among the Bedouin population in the Negev.

While in 2011 genetic carrier testing was done at the genetics institute at Soroka Medical Center, the outreach program was effectively implemented in 2012, making nurse-mediated carrier testing services available the community clinics. We analyzed differences between various parameters of the cohort tested in the community in the year 2012 and the cohort tested at the genetic counseling clinic in Soroka Medical Center in 2011; we examined what are the factors that influence the decision to carry out or refuse to perform the carrier testing, and to undergo medical diagnostic prenatal tests.

Research objectives

- 1. To assess the efficiency of accessibility of the community genetic services and their impact on uptake of genetic carrier screening in community health clinics among the Bedouin population in the Negev in the year 2012, compared to the provision of services only in Genetics Institute (2011).
- 2. To examine the factors affecting compliance for further genetic counseling and testing when both spouses are carriers (genetic counseling at the Genetic Institute, performing prenatal diagnosis through chorionic villus sampling / amniocentesis) and the resolutions adopted following the diagnosis of a disease.

Methods

The study population: All Bedouin visitors in the community nursing genetic services (CNGS) in 2012 and all Bedouins attending genetic carrier testing services at the Genetics Institute at Soroka Medical Center in 2011.

Data were analyzed using SPSS version 26.0. Univariate analyses include t-tests for comparison of means (or Mann-Whitney tests as appropriate) for numerical variables and chi-square tests for nominal variables.

P-Values were calculated (significance determined at p<0.05).Correlationsbetweenthevariables(Pearsonandspearmen, by type of variables) were examined to ensure no co-linearity.

Results

Most of the women who carried out the test for HRD were younger (average age 26.2) compared with those who did not (average age 27.8). The average number of pregnancies of women who did the test was 3.5 compared to women who did not (average 4.5 pregnancies per woman). The average number of children was 3 children for women who underwent testing as compared with an average of 3.9 children per woman for those that chose not to be tested. The majority of women who came to CNGS in 2012 were pregnant women (93.1%). The average week of pregnancy, number of children and number of pregnancies were 1,3, and 18 respectively. With the introduction of the community outreach program there was an increase in the percentage of women who carried out the tests: table1.

Uptake of recommended genetic screening tests include HDR by the years 2011-2012

Variable	2011	2012	P- value	
	N (%)	N (%)		
Women gave birth	7768	8379		
* Number of tests done	1221	1965		
Women uptake HRD test	1164 (14.9)	1871 (22.3)	<0.001	

* Women could have been tested more then one in a calendar year and could do the test before or during pregnancy or after delivery/

Results of prenatal tests (CVS/Amnio) done by the years 2011-2012 and decision made after that

	2011		2012		
Variable	Total	Тор	Total	Тор	
	N (%)	N (%)	N (%)	N (%)	
CVS:	28		43		
Healthy fetus	7 (25.0)		17 (39)		
Carrier fetus	14 (50.0)		14 (31.0)		
Affected fetus	7 (25.0)	5 (71.0)	12 (30.0)	10 (83.3)	
Amniocentesis:	14		9		
Healthy fetus	4 (28.5)		5 (56.0)		
Carrier fetus	7 (50.0)		2 (22.0)		
Affected fetus	3 (21.5)	1 (33.3)	2 (22.0)	1 (50.0)	

Consanguinity distribution in the study population in the year 2012 (N = 1754)

Variable	N (%)		
Consanguinity	1081 (61.6)		
Of them:			
First & Double cousin	595 (33.9)		
Second cousin once removed	131 (7.5)		
From the same tribe	355 (20.2)		
Not related	673 (38.4)		

Factors associated with uptake of genetic screening test - results from a multivariate logistic regression analysis

Variable	P- value	OR	95% CI	
Knowing Hebrew language (y/n)	<0.001	2.22	1.39	3.56
Gestational age at visit (weeks)	<0.001	2.26	1.64	3.12
Family history of genetic disease (y/n)	0.01	2.46	1.19	5.07
Explanation given by the referring (y/n)	<0.001	2.75	1.87	4.05
Pregnancy number	0.008	1.89	1.1	3.08

Only 10.8% of the women refused to perform a genetic screening test. Of those women attending the CNGS in 2012, 44.6% completed primary school education, only 10.1% were employed. For most of those women (81.8%) it was their first visit to CNGS. Most came alone, and 59.9% were followed regularly during their pregnancy by a gynecologist in a primary health clinic. Of the women attending the CNGS in 39.1% 2012, were referred for genetic counseling at Soroka Medical Center – only about half of them actually complied and attended the genetic counseling. The main reasons for referral were family history of genetic disease and problems detected in the testing during pregnancy.

Conclusions

The study highlights the efficacy and importance of: - making genetic services available to the public in rural areas; - promoting specialized nursing expertise/ services in genetics, targeted at specific population at risk; - enhancing genetics aspects in nursing academic programs, enhancing public health and awareness in genetics-related issues.

Keywords: Prenatal genetic screening, prenatal diagnosis, culture, the Bedouin sector, genetic diseases, inbred, polygamy, making a decision, genetics community.

