Distribution of Hemoglobinopathy Disorders in Saudi Arabia Based on Data from the Premarital Screening and Genetic Counseling Program, 2011-2015

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Introduction

- Hemoglobinopathy disorders are genetic defects in hemoglobin
- High mortality and morbidity rates present in thalassemia disorder (β-thal) and sickle cell disease (SCD)
- Higher prevalence of β-thal and SCD in Saudi Arabia compared to neighboring countries
  - In 2011, 0.05% β-thal and 4.5% SCD prevalence
- Saudi Arabia initiated a mandatory premarital screening and genetic counseling program (PMSGC) in 2004
- Trends in prevalence rates of β-thal and SCD have not been examined since 2011

Objectives

- Assess recent trends in β-thal and SCD and their distribution by demographic characteristics and geographic regions using data from the PMSGC program

Methods

- Secondary data analysis
- 1,230,582 individuals
- Data obtained from Department of Genetics of the Saudi Ministry of Health (MoH), PMSGC program
- Included all couples within 13 administrative regions from February 2011 to December 2015
- Status of β-thal and SCD categorized as positive, negative, and carrier
- Prevalence rate and 95% (CI) of β-thal and SCD estimated by study year and geographic region

Results

Population characteristics: 49.7% men; average age 27.8 years (SD 8.85 years) and 50.34% women; average age 22.6 years (SD 6.4)

Table 1. Overall prevalence rate (per 1000) for β-thalassemia and Sickle Cell disorders in all region, Saudi Arabia, 2011–2015

	Disease	Trait	Total
β-thal	0.7	12.9	13.6
SCD	3.8	45.8	49.6

β-thal major: PR (per 1000) ranged from 1 in 2011 to 1.6 in 2015.

β-thal trait: PR(per 1000) ranged from 24.2 in 2011 to 12 in 2015.

SCD: PR (per 1000) ranged from 5.2 in 2011 to 3.4 in 2015.

SCT: PR (per 1000) ranged from 37.1 in 2011 to 46.7 in 2015.

Table 2. Prevalence rates (PR per 1000) for β-thalassemia Trait and Sickle Cell Trait in Saudi Arabia form 2011 to 2015

	Population screened	B-thalassemia Trait			Sickle Cell Trait		
Year	1,230,582	Positive test	PR	Confidence Interval	Positive test	PR	Confidence Interval
2011	78072	1892	24.2*	23.2-25.3	3304	42.3	41.0-43.8
2012	258581	4557	17.7*	17.1-18.2	13055	50.5	49.7-51.3
2013	268097	2837	10.6*	10.2-11.0	13406	50.0	49.2-50.8
2014	276236	2666	9.7*	9.3-10.0	13490	48.9	48.0-49.7
2015	305871	3657	12.0*	11.6-12.4	15307	50.0	49.3-50.8

Figure 1. Prevalnce Rates* of β-thalassemia among differnt regions during years, Kingdom of Saudi Arabia, 2011 – 2015*

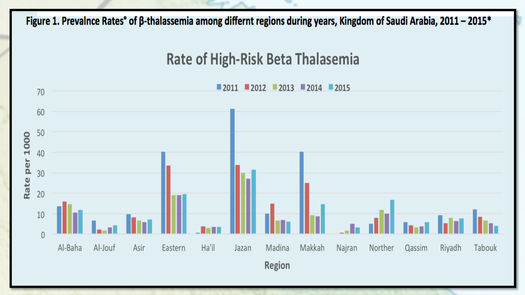

Figure 2. Incidence Rates* of High risk presention of Sikle cell disease among differnt regions during years, Kingdom of Saudi Arabia, 2011 – 2015

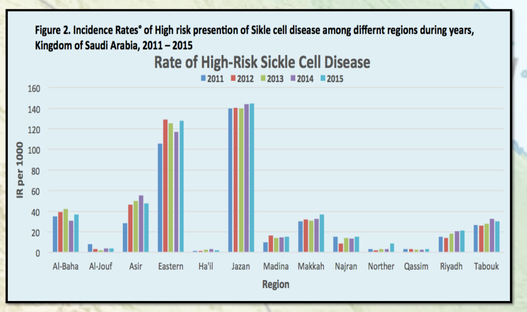

Table 3. Prevalence rate for β-thal and sickle cell traits and disorders in regions with highest burden

	Jazan Region Prevalence rate (95% Confidence Interval)	Eastern region Prevalence rate (95% Confidence interval)
β-thal trait	32.1 (30.8-33.4)	23.7 (23.1-24.4)
β-thal major	0.6 (0.5-0.8)	0.4 (0.3-0.5)
SCT	135.7 (133.2-138.2)	114.4 (113.0-115.8)
SCD	6.8 (6.2-7.4)	9.8 (9.4-10.2)

Conclusion

- Over a 5-year period, a decreasing trend in the prevalence of β-thal was observed
- SCD rates were rather stable over time
- Compared to 2004–2009 rates:
  - β-thal major rates were similar in 2011–2015
  - β-thal carrier rates decreased (from 32 to 13 per 1000)
  - SCD and SCT rates similar
- Highest prevalence observed in Eastern region and Jazan region, similar to distribution reported in 2004 – 2011 study
- Program is moving towards reaching goals of lowering prevalence of β-thal.

Recommendations

- Rates still especially high among certain regions like Eastern Region and Jazan; these regions should be targeted with intensified awareness programs
- β-thal and SCD rates in newborns need to be assessed to evaluate program effectiveness
- More effective genetic counseling programs for at-risk couples
- Awareness programs among youth about risks and recepetivity to genetic counseling

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