

# TIF conference presentation in detail

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## Abstract

The factors determining the health behaviours may be seen in various contexts: physical, socio-economic, cultural and political. So the utilization of a health care system, public or private, formal or non-formal, may depend on socio-demographic factors, social structures, level of education, cultural beliefs and practices, gender discrimination, status of women, economic and political systems environmental conditions, and the disease pattern and health care system itself. Policy makers need to understand the drivers of health seeking behaviour of the population in an increasingly pluralistic health care system. Also a more concerted effort is required for designing behavioural health promotion campaigns through inter-sectoral collaboration focusing more on disadvantaged segments of the population.

Thalassaemia is the most prevalent genetic blood disorder in Pakistan. It is estimated that there are 8-10 million Thalassaemia Minor cases in the country with a prevalence of 5-6%. It is also estimated that about 100,000 patients suffering from Thalassaemia Major exist in Pakistan and every year this number is increasing by about 6,000. Pakistan is witnessing this large increase in thalassaemic patients due to a lack of proper coordinated, nationwide efforts to contain the inherited form of anaemia, and general public awareness.

Different research studies and diagnosis services are carried out in Pakistan on Thalassaemia prevalence. One such service for prenatal diagnosis of  $\beta$ -thalassaemia was introduced in Pakistan in May 1994. Two renowned Islamic scholars, consulted before the service was introduced, ruled that a pregnancy can be terminated if the fetus is affected by a serious genetic disorder, and if termination is before 120 days (17 weeks) of gestation. During the first 3½ years of the service 300 couples requested the test. Almost all the couples had been informed by their treating doctors. Most diagnoses were made between 10 and 16 weeks of gestation, and only 15 (5%) were reached after the 16th week. DNA analysis was by the amplification refractory mutation system (ARMS). A multiplex ARMS was developed in which three primer combinations identified the mutations in 91.5% of the couples. In 13 couples (4.3%) linkage analysis was required for the fetal diagnosis. In 47/53 (88.7%) women carrying an affected fetus the pregnancy was terminated. In six cases it was declined principally on religious grounds. Postnatal confirmation of the prenatal diagnosis was possible in 117 unaffected children. One year after the start of the

service, interviews with 141 couples with an affected child showed that 72% knew of the availability of prenatal diagnosis. Thirty-two of the informed couples had had a pregnancy, but only 18 (56%) used prenatal diagnosis. The main reasons for non-utilization of prenatal diagnosis were the cost of the test and fear of undergoing the test, though some gave no clear explanation. This study demonstrates that prenatal diagnosis is feasible and acceptable in a Muslim country such as Pakistan (Shoab, Salim 2000).

Another study on Pakistan characterized 1216 beta-thalassaemia alleles from the five major ethnic groups of the country. The complete spectrum comprised 19 different mutations. There are important ethnic and regional differences in the prevalence of mutations. The five most common mutations, IVSI-5 (G-C) (37.3%), Fr 8-9 (+G) (25.9%), del 619 (7.0%), Fr 41-42 (-TTCT) (6.7%) and IVSI-1 (G-T) (5.4%), constitute 82.3% of the total. Fr 8-9 (+G) is the most common mutation in Northern Pakistan (41.3%), whereas IVSI-5 (G-C) is the most frequent mutation in Southern Pakistan (52.2%). Six subjects with transfusion-dependent thalassaemia major showed only a single mutant allele. One subject with transfusion-dependent thalassaemia major showed a novel 17 bp deletion involving Cd126-131. Our findings provide a comprehensive basis for carrying out prenatal diagnosis of thalassaemia in a geographical area where it is found in high frequency (Ahmad, Saleem 1996).

## Brief country profile of pakistan

The Indus Valley civilization, one of the oldest in the world and dating back at least 5,000 years, spread over much of what is presently Pakistan. During the second millennium B.C, remnants of this culture fused with the migrating Indo-Aryan peoples. The area underwent successive invasions in subsequent centuries from the Persians, Greeks, Scythians, Arabs (who brought Islam), Afghans, and Turks. The Mughal Empire flourished in the 16th and 17th centuries; the British came to dominate the region in the 18th century.

The separation in 1947 of British India into the Muslim State of Pakistan and largely Hindu India.

### South Asia

Total Area: 796,095 sq km

Land: 770,875 sq km

Water: 25,220 sq km

Population: 208 million (2017)

Country comparison to the world: 36

### Land boundaries

Bordering the Arabian Sea, between India on the East and Iran and Afghanistan on the West and China in the North.

### Total bordering Area

6,774 km, with Afghanistan 2,430 km, with China 523 km, with India 2,912 km and with Iran 909 km, Coast line 1,046 km.

It also controls Khyber Pass and Bolan Pass, traditional invasion routes between Central Asia and the Indian Subcontinent.

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