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Premarital Screening of Thalassemia in Punjab: A Policy Perspective

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Abstract

Thalassemia is one of the most prevalent inherited haemoglobin disorders worldwide, with a particularly high burden in low- and middle-income countries such as Pakistan. With a carrier rate of approximately 5–8% and thousands of affected births annually, β-thalassemia poses significant challenges. Premarital screening has been proposed as a preventive strategy; however, policy development and implementation remain limited in Punjab, Pakistan.
This study aimed to explore the political, institutional, legal, and socio-cultural challenges associated with the formulation and implementation of a premarital thalassemia screening policy in Punjab, and to examine stakeholders' perspectives regarding the proposed Thalassemia Prevention Act.
A qualitative research design was employed using an interpretive approach. Purposive sampling was used to recruit key stakeholders involved in health legislation and policy formulation, including policymakers, legislators, bureaucrats, and health program officials. Data were collected through in-depth semi-structured interviews. Thematic analysis was applied to identify major themes and subthemes related to barriers in policy formulation and implementation.
Findings revealed multiple interrelated barriers to premarital screening policy development. Key challenges included limited evidence-based policymaking, inadequate research organization and interpretation, insufficient bureaucratic engagement during early formulation stages, and lack of detailed legal enforcement mechanisms. Political will emerged as a critical enabling factor, although inconsistencies in leadership commitment delayed legislative progress. Socio-cultural factors, including high rates of consanguineous marriages and religious sensitivities surrounding antenatal diagnosis, further influenced policy feasibility. Economic analysis indicated that preventive screening could significantly reduce long-term treatment costs associated with thalassemia major. Participants emphasized the importance of public awareness campaigns, stakeholder collaboration, and integration of scientific research into legislative processes.
The successful implementation of premarital thalassemia screening in Punjab requires sustained political commitment, institutional coordination, robust legal frameworks, and evidence-informed policymaking. A comprehensive and culturally sensitive policy approach

could shift Pakistan's response from reactive disease management to proactive genetic prevention, offering a sustainable model for other low- and middle-income settings.

Keywords: Thalassemia, Premarital Screening, Health Policy, Evidence-Based Policymaking, Genetic Disorders, Pakistan, Public Health Governance

Introduction

Over the past few years, genetic diseases have significantly transformed the overall disease pattern worldwide. This scenario has led to an escalation in the global burden of disease and become a challenge for public health in recent times.

Haemoglobinopathies, such as thalassemia and sickle cell disease, have been prevalent. They have been on the front line over the years to provide public health professionals with a track to think about and work on. Thalassemia is an inherited autosomal recessive hematologic disorder that can lead to anaemia. It is caused due to imbalances in globin chains, i.e. the decreased ($beta^{+}$) or absent ($beta^{0}$) synthesis of one or more globin chains (Rund & Rachmilewitz, 2005). The severity of thalassemia depends on the extent of disturbances between the globin chains.

Thalassemia is one of the most common single gene disorders globally. However, these disorders are most prevalent in low and middle-income countries (Kattamis et al., 2020; Weatherall et al., 2006). The percentage of carriers of haemoglobin disorders like Thalassemia, sickle cell anaemia and haemoglobin E is 7% worldwide, i.e. approximately 300,000-500,000 children are born with these diseases every year, the majority of which belong to developing countries (Weatherall et al., 2006). Out of this prevalence, 1.5% population is the carrier of beta-thalassemia (Colah et al., 2010).

Pakistan is facing the hazards of this common disorder. Although considerable statistical data about β-thal is not available, literature states that approximately 4000-9000 patients are born with thalassemia major in Pakistan. This huge number in a country like Pakistan is a great concern for the health sector and public health professionals (Ghafoor et al., 2021; Khateeb et al., 2000). In the Eastern Mediterranean Region, Pakistan has 5-7% of thalassemia carriers, meaning 9.8 million of the total population (Khaliq, 2022; Ansari et al., 2011).

A high risk of mortality and morbidity is associated with β-thalassemia. Rund & Rachmilewitz, (2005) state that as successful bone marrow transplantation is not available, children who get a blood transfusion in early childhood have improved quality of life, but iron chelation therapy is frequently needed. Iron chelation therapy with deferoxamine is an expensive procedure. It costs $4400 per patient and treating a cohort of affected children annually will cost 90,000 units of blood and $22 million for deferoxamine. This estimates to be more than 4% of the total health expenditure (Ansari et al., 2011). In this way, a major part of health expenditure will be spent on thalassemia management, which is a cause of great concern for low-income countries like Pakistan, where health has only a 1.2% share of the Gross Domestic Product. (Pakistan Economic Survey, 2020-21).

Antenatal screening and pre-implantation diagnosis were one of the suggestions put forth by health authorities, but post diagnosis options were difficult to opt for as abortion is not acceptable by most communities, religions and individuals as well (Rahman & Lodhi, 2004). When this situation exists the premarital screening program becomes a desirable but challenging initiative (Shariq et al., 2014).

Pakistan is a low-income country with a population of approximately 225 million (World Bank, 2021), having various health burdens. Consanguineous marriages are permissible in Islam, and Pakistan being a Muslim country, widely practises consanguinity. This can lead to an increase in the prevalence of genetic disorders (Kamil et al., 2021; Maryam & Alam, 2025). The carrier rate of beta thalassemia is 5-8% in Pakistan (Ansari et al., 2011). There is no premarital carrier-screening program for thalassemia, but antenatal screening is offered in various parts of the country. If fetal anomalies are detected, abortion is the only option for couples, but it is not legally allowed.

The current burden of thalassemia in Pakistan underscores the urgent need for effective and sustainable prevention programs. Premarital screening for prospective couples and youth to be married is a feasible option to help reduce the disease burden by stopping at-risk marriages and defective births. Nevertheless, there are some challenges related to this type of prevention program but public educational programs, awareness and advocacy can help manage the obstacle to premarital screening practices.

Premarital screening has been a policy debate in Punjab, and the Provincial Ministry of Health and spokespersons have presented a bill and tried to implement the policy for the carrier screening of thalassemia. However, there are certain barriers and challenges that are faced by legislators and policymakers. The present study aims to identify the challenges and views of stakeholders of premarital screening policy from the supply side and suggest a framework for the execution of failure free policy.

The challenges for health screening policies for either genetic disorders, neonatal screening or screening of sexually transmitted diseases are more or less similar in social settings of countries like Pakistan. Firstly, there is a lack of policy for such screening programs, and if the policy is formulated, the scientific aspect is overlooked during the policy formation. Lack of infrastructure and equipment is another challenge for the suitable provision of the screening program. Moreover, socioeconomic, and cultural constraints and deficiencies in the community healthcare system are also key hurdles for screening programs. In addition, the lack of human resources professionals and awareness among the public are major factors in the hindrance of screening policy execution (Mumtaz et al., 2019).

Thalassemia is a prevalent genetic disorder in Pakistan, treatment of which is challenging for the affected families as well as the health care providers. Although premarital screening has its shortcomings and detriments, it is still proven to be an effective method for preventing hereditary diseases.

The present study aims to identify the challenges and barriers to premarital screening based on the views and perspectives of policymakers and legislators who are the key stakeholders for policymaking and its enforcement. The study also aims to explore the major causes of delay in the Thalassemia Prevention Act pertaining to premarital screening of thalassemia. The current study is also focused to explore the legislative and policy challenges. All the dimensions related to policy making, from formulation to implementation are explored in the study. The research questions are developed according to the study domain of the research and major stakeholders are involved to gain the deeper view of prospects related to premarital screening of thalassemia in Punjab.

Research Question

What are the legislative and policy challenges perceived by legislators and policymakers regarding mandatory premarital thalassemia screening?

Methodology

This study explores the challenges and barriers related to the policy formulation and implementation of premarital screening by using qualitative research method. There are various challenges that policy makers face in the form of lack of awareness, infrastructure and public opposition. In order to deeply understand the meanings attached to experiences and perspectives of policy makers and legislators, researcher explores it through the views of those directly related to the policy of premarital screening.

The qualitative research model is used because the present research interprets the social actions and sense-making of the meanings attached to the professional views of key stakeholders involved in constructing a phenomenon. The professional views of legislators and policy makers are different in the context of this study so the study uses the ontological stance of relativism. According to the current study, the relativism explains the phenomenon of challenges and barriers i.e. what hinders the formulation and implementation of premarital screening policy. While epistemological stance of interpretivism explore the professional views and perspectives of stakeholders related to pre marital screening of thalassemia.

Table 1: Philosophical underpinnings of the research paradigm (Crotty, 1998)

	Ontology ***(What Is?)***	**Epistemology** ***(How we know what we know?)***	**Methodology** ***(Strategy or Plan of Action)***	**Method** ***(Techniques or Procedures)***
Philosophical Underpinnings	Relativism	Interpretivism	Research Methodology	Tools
According to the Study	• Challenges & Barriers • Prospects	Professional Views	Qualitative Research Method	Interviews

The researcher interprets the prospects and views along with the meanings attached to them, among policy makers. Understanding social reality constructed due to the implementation of new health prevention programs like premarital screening of thalassemia falls under the domain of an interpretive approach rather than a positive thought. This approach considers how a particular health care phenomenon is created and how it influences the prospects of people directly related to the initiation and provision of health care programs and vice versa. This means that how the views are created and attaching meaning to these views in a specific context is of interest to the qualitative researcher. So, the framework of this research, which includes the research design, data collection method, data analysis and analytical process, is based on how the professional views of stakeholders construct the reality of policy dynamics.

Sampling

Data was collected using purposive sampling techniques in qualitative research. Purposive sampling means that the researcher intentionally selected those respondents who can fulfil the purpose of the study by giving in-depth information. Fifteen policy makers and legislators were interviewed for this study. For the recruitment of policy makers, initial identification was done through the provincial health minister, provincial health department

and personal contacts. The Provincial Health Minister was contacted using personal and professional contacts. After understanding the objectives of the study, he helped in contacting the other policy makers who could be helpful in providing the required information. As the study objectives were to explore the challenges of premarital screening from the supply side, i.e. policy making, the recruitment of participants was carefully made, keeping in view that all the major stakeholders related to thalassemia prevention policy be included. The respondents who are directly related to the health legislation and policy making of thalassemia prevention are involved. Special consideration is given to the selection of respondents so that none of the policy dimensions remains undiscovered. Participants were selected from all the domains starting from policy preparation to formulation, implementation, budgeting, and recruitment for human resource.

Professional demographics of study participants

The policy makers and legislators who participated in the study were all working at significant administrative posts. The policy makers among the bureaucrats have passed the special services (CSS) examination and got special training for recruitment of administrative and policy related posts. They all had more than 15 years of work experience in the policy related fields. The legislators, were the elected members of the provincial assembly and had long standing affiliation with their respective political party. They were well aware of the legislative procedure and their professional experience helped the researcher to gain insight into legislative aspects and challenges associated with introducing pre marital screening policy for thalassemia.

Development of Interview Guide

The development of interview guides needs a rigorous effort so that the aim of the study is fulfilled, maintaining the well-being of the respondents. The researcher opted for empirical data and extensive literature to get a holistic view of the research questions. A semi-structured interview guide was used for data collection and in-depth interviews were conducted according to research rules. In qualitative research, in-depth interviews are referred to as non-directive and open-ended discussions between the researcher and respondents to gain information about the perspectives of experiences and views of participants about a phenomenon or situation (Taylor et al., 2015).

Based on the epistemological grounds of the study, researcher used an unstructured form of interviews. These types of interviews have a flexible nature, they facilitated the researcher to spend more time with the participants and explore their views more deeply. As the project proceeded, more themes were added and explored that were not considered in the initial stages. Some probing questions were also added to the interview guide to get a deeper understanding of their perspective.

Composition of the interview guide

The first part of the interview guide was based on general information and the demographic profile of the study participants. Secondly, open-ended questions were asked from respondents about the topic for deeper investigation. Finally, respondents were asked to give recommendations to get their valuable views and make the detachment process easy. The guides were prepared with understandable, short and clear questions in a logical manner so that maximum information could be gathered.

Tools for data collection

Interview guide was prepared for the policymakers regarding the policy challenges of pre-marital screening. In this guide questions were asked about the general process of health

legislation. It proceeded with inquiries about the views of study participants regarding premarital screening of thalassemia. Further, specific challenges related to premarital screening policy of thalassemia were discussed.

The interviews were conducted in the local language Urdu and the guides were also translated into Urdu. The interviews were audio recorded. Two audio recording devices were used to record the interviews so that if one device accidentally loses the data the other may be used. The recorded interviews were immediately transferred to the researcher's laptop and storage disks to keep the data safe in case of any technical problem.

Positionality and Stand Point

The present study involves the professional difference in the context of policy making as the researcher was public health professional related to the field of health care and public health academia. Investigating the challenges of legislation and policy making certainly brought professional divergence in this context. The reflexivity helped to negotiate the professional discrepancies in the scenario of the present study.

The study follows the feminist standpoint epistemology on the grounds that enable the researcher to minimise power differentials. The differences occur on the basis of gender, education, social class, profession, etc. The difference due to professional level occurs because the respondent were in a privileged professional position. In the context of the present study the researcher carefully worked for minimising the professional level difference in case of legislators and policy makers. Although the educational level of the researcher was not different from that of the participants, the privileged social and professional position subjected them to be in a different matrix than the researcher.

At times in the legislative setting, the researcher was seen as an outsider trying to intervene in the legislative process and policy-political affairs of the institution. This could restrict the participants from sharing the actual information and details about the difficulties of the policymaking process. The researcher was able to mitigate these issues to some extent and continuously assured them that the information provided by the participants will only be used for the research purposes. Another strategy used by the researcher to overcome the power differentials was keeping updated about the political situations. It helped the researcher in such a way that at times during the interviews when the political barriers were discussed the researcher was able to give her viewpoint in that context. This assured the participants that the interviewee, even being a member of the public, is well aware of the political challenges and scenarios so that they could freely share their perspectives, and the researcher diligently extracted the themes from that discussion. The researcher also provided her identity so that sharing the information can become easy for the respondents.

Moreover, the researcher's gender was another issue that made the data collection method difficult. Among the policymakers and legislators, all the respondents were males, as females do not take much part in politics, neither are they encouraged to do so. So, visiting the offices of legislators was not an easy task. For this purpose male members of the research team accompanied the researcher to collect the data.

Data Analysis

After the completion of field work, the interviews were transcribed and translated from Urdu to English. Key themes and sub themes emerged for data analysis and interpretation of the findings, through the process of coding. The identities of the participants were kept confidential and names and designations were changed and replaced with codes for further analysis. Only the domain to which the participants belonged were used i.e. legislation

or policy making, in order to ensure their anonymity. Findings of the study were written after the analysis and the important quotes relevant to the themes were used in write up.

Transcribing and Analysing Data

Positioning analysis in grounded theory approach

The data analysis of the present study positions itself on the principles of grounded theory, and the researcher has attempted to develop the theory during the process of data analysis while comparing the themes that emerged at every stage. Analytical coding and memo writing for elaborating the categories and their relationship is done according to the grounded theory methodology. Purposive sampling is done to construct a newly emerged theory through data collection and analysis. In the process of data collection already existing theories are not compared for the deduction of results but an independent data analysis is done to create an idea based on the findings of data.

The themes that emerged in the study are discussed in the literature with reference to other diseases in similar healthcare settings. This helped investigate the reliability of findings in the context of thalassemia premarital screening in Punjab. The study findings in the form of themes that emerge from the data analysis are discussed in detail in the findings section.

Analysis Strategies

All the interviews were recorded on a voice recording device. Besides recording the interviews the researcher also took field notes. The interviews were recorded with the permission of the participants and minute details observed in the field were observed and noted in the field notes that helped the researcher in analysing data and writing up the stage. After conducting each interview, the researcher listened to it on the same day to get an idea and holistic picture of the discussion she had with the participant.

After completing the fieldwork, all the interviews were listened to in detail, and the researcher transcribed the interview for the sake of getting an idea about the coding and themes that are to be discussed while writing the research findings. In the next step, the researcher coded and translated the interviews from Urdu to English. Key themes and subthemes emerged during the process of coding and translating. The study findings consist of two chapters. One chapter explains the prospects of legislators and policymakers, while the other chapter discusses the challenges and barriers of service delivery.

Transcribing and translating

Transcribing the research data is not a simple task, it is not only about giving codes and extracting themes. It involves a clear understanding of the related problems such as body language and the way of talking of the interviewee. The transcribing process itself starts the data analysis. During the interviews in participants' offices, the researcher kept noting the observations and field notes. While transcribing, analysing and writing up, the researcher referred to the field notes. It helped her to more clearly understand the emerging themes and kept her in a continuous imaginary dialogue with the respondent. The researcher adopted the technique of profoundly immersing into the data while transcribing and translating the interviews. Qualitative data analysis is a time taking and laborious task. So, it took long hours to develop a verbatim transcript for a one hour interview.

Coding of empirical data

After transcribing, the next step was the coding of data. In qualitative data analysis coding of data is the major step that moves the research forward towards the finding the results. The identification of codes was highlighted during the phase of data collection, which later on emerged as codes during the analysis phase. The inductive codes that emerged from the original data were organised according to the deductive codes emerging from the

literature. These codes were finalized based on the similar patterns that existed between the inductive and deductive codes. Codes and their description were noted carefully for consistency in the results of the present research.

Themes and categories

Key themes and sub themes emerged from the data, and were analysed based on emerging themes. The categories showing similar descriptions were organised under a single theme to provide a clear and coherent view of the study findings. These themes explained the challenges and prospects of legislators, policymakers and health care providers. Organising the categories and themes helped to refine the analysis process and shaping the study conclusions. The names of the participants and their designations were changed and codes were given for further analysis. In the written thesis, only the names of specialized departments were used to which the participants belong were used to ensure anonymity. Similarly, alternative wording was used instead of designations to hide their identity in connection to their designations, departments and ministry to which they belonged. The finding, after analysing the data, were written and the important quotes related to themes were used in the write- up.

Seeking Rigor and Trustworthiness

Rigour and trustworthiness in qualitative research provide the research with accuracy and consistency, especially in the analysis and interpretation phase. Qualitative rigor enables the researcher to develop the trustworthiness in the generalisability of the qualitative findings (Thomas & Magilvy, 2011). In the current study the researcher used various techniques for seeking rigor and trustworthiness. The subjectivity of researcher and respondents in acknowledged by qualitative research but it is important to ensure that the subjectivity does not interfere with the research findings. Some primary techniques are discussed at the analysis and interpretation stage.

Triangulation of Data Sources

Various rigorous techniques for enhancing the quality of qualitative data analysis are used. Triangulation of data sources means, using all the data sources used in the study to maintain consistency and gain a comprehensive understanding. Moreover, it is also used to test the validity of the data sources used (Carter et al., 2014). In the present study, the researchers used all the data sources, i.e., in-depth interviews with legislators and policy makers. Moreover, they included the data from the records of the thalassemia prevention program for the purpose of triangulation. This resulted in cross validation of the data to develop an authentic and valid interpretation.

Reflexive Journaling

Reflexive journaling is another technique for maintaining rigour and trustworthiness in data analysis of qualitative research. It helps facilitate reflexivity throughout the research process right from the beginning and clarifies the personal subjectivity by posing questions to subjectivity (Ortlipp, 2008; Creswell & Miller, 2000). During the present study, from planning and designing the research and through field work stages to data analysis, the researchers maintained the reflexing journals. These journals were used during data collection to note personal observations. During the process of data analysis and developing codes and categories, the researcher referred back to the reflexive journals to gain a better understanding of data and its contextualization.

Thick Description: Contextualization of Qualitative Data

After the coding was carried out, each code was examined thoroughly for the purpose of carrying out a thick description. Thick description helped the researcher to understand each code through the views of stakeholders and getting a clear picture in terms of patterns of

behaviours and experiences of legislators and policy makers. The thick description aimed to examine the views of study participants in the context of legislation, health policy. The experiences, interactions and professional behaviours interpreted the meanings through thick description for the process of further analysis of data that includes categorisation and theme development.

Referential Adequacy

To gain the trustworthiness in data and support the data analysis, referential adequacy technique is used. Referential adequacy ensures that the basis of conclusions and findings of the study are grounded in the data. It describes data analysis through holistic and rich contextual background data (Byrne, 2001). The researcher applied the technique of referential adequacy and ensured that the interpretations and conclusions about the legislative system and policymaking originally lie in the data. This ensured that the data is analysed and results are drawn based on views of study participants and not the subjectivity of the researcher being an insider to the study settings.

Member Check: Participant Validation

When the researcher interpreted and analysed the data in this study, it was returned to participants for validation. The researcher checked the results through some of the legislators, and policymakers to make sure that the findings of the study were accurate and made sense. The results were consistent in terms of conclusion, researcher's interpretation and participants' validation. The write up of the analysis sectiom was done after the member check.

Ethical Approval

The current study is a qualitative study based on the views and experiences of policy stakeholders. There was no human harm (physical, psychological or emotional), therefore ethical approval from the institutional ethical review committee was not required. However, the written and verbal informed consent was obtained from the participants and all the participants were ensured that the data collected will be used only for research purposes. Moreover, the respondents included legislators and political figure, so the names and designations of all the respondents were anonymised and data was kept confidential.

Findings

The findings look into various challenges and barriers related to policy formulation and legislation as well as the prospects of major stakeholders of policymaking. The legislators and policymakers involved in the drafting and preparation of the initial premarital screening bill for thalassemia in 2018 are involved in this study. This bill was named as Punjab Prevention and Control of Thalassemia Act. These stakeholders are well aware of the challenges and barriers faced by health policies in the legislative, formulation and implementation phase. Keeping in view the delay in the premarital screening act of thalassemia and its challenges in the dimensions of legislation, formulation and future implementation are covered by the researcher. This gives a holistic view of the barriers that health policies generally face and particularly those the premarital screening policy is going through.

The challenges of premarital screening of thalassemia are discussed in the domains of policy making and legislation. The researcher interviewed the stakeholders who were mainly involved in the initial preparation of the screening bill. They include the health care providers from the Punjab Thalassemia Prevention Program, legislators who are also members of the provincial assembly and ministers of both wings of the health department, i.e. "Primary and Secondary Health" and "Specialised Health and Education". The policy related and legislative

barriers are discussed in terms of political, economic and public related barriers. As the study moves forward, it is determined that health policies generally face many difficulties in the formulation and implementation phase while the premarital screening policy is still at the very initial stage of preparation and legislation has not been done. So, the scenario for this particular prevention policy is different.

Furthermore, the views of stakeholders about the initiation and presentation of the bill and implementation of the policy afterwards provided a holistic view. It explains the benefits and risks of thalassemia prevention in Punjab. Finally, the chapter moves towards the professional views of stakeholders about the premarital screening of thalassemia as well as the prerequisites to overcome challenges in the way of Punjab Prevention and Control of Thalassemia Act.

Political Barriers

Political leadership plays an important role in policy development as well as law making. When policies are developed the skills and leadership of political stakeholders among the policymakers is needed. Public policies, including health policies are raised on foundational grounds which depend on several significant factors. Like all existing laws and policies, the Punjab Thalassemia Prevention Act on premarital screening of thalassemia is in the process of formulation and legislation. Present study attempts to find the major political barriers in the ways of implementation of premarital screening act. Moreover, the views of stakeholders and policymakers are explored to correlate the challenges with their prospects.

Barriers to Policy Formulation

Policy formulation is the first major step after planning and designing policy. Formulation of a policy includes putting each step of the agenda on the program planned and set by the government (Jann & Wegreich, 2007). Policy formulation is a crucial phase during which things are settled by those who are directly involved in the initiation and legislative process. During the formulation phase, various factors can cause hindrances in the process of active formulation. These factors are discussed in the form of themes in this study, and the respondents shared their views about the factors that are the major barriers in the formulation of premarital screening policy as well as discussed the requirements of policy formulation. The prospects of the respondents about the requirements of the policy formulation emerged in the form of the following themes.

Table 2: Sub themes and codes in the theme of Barriers to Policy Formulation

Theme	Sub Theme	Codes
Barriers to Policy Formulation	Protocol and guidelines for policy making	• Basic requirement for policy formulation • Guidelines already present • Well defined policy protocols result in well formulated policy • Not a challenge for premarital screening policy
	Evidence based policy making	• Scientific evidence is needed • Evidence based policy making approach is needed • inform policy decisions, create a knowledgeable

Theme	Sub Theme	Codes
		environment, reduce uncertainty in policy decisions and eliminate the political divide
	Data organisation	• Research is not policy focused • Scientific data is not organised according to policy approach • Lack of scientific research in the field of genetic diseases and prevention
	Interpretation of evidence	• Evidence informed policy decision • Scientific background can be understood • Policy alternatives are suggested • Lack of policy focused scientific interpretation
	Bureaucracy and policy making	• Lack of bureaucratic partnership • Less involvement in policy making • Non-involvement in the initial phase of thalassemia prevention policy

When policymakers follow the prior templates: Protocol and guidelines for policy making

Protocols for policymaking are a basic requirement for the formulation of policy. Previously many action policies have been formulated with their guidelines and protocols for health reforms on the national level. Presently in the fresh decentralised system, health is the responsibility of provinces and the ministry of health has to plan and take actions to improve the healthcare system and health service delivery. The guidelines and protocols for the policy formulation are already present, and commitment is needed for a well-formulated policy to be executed. Collaboration, commitment, communication, coordination, and context are the main areas that need to be focused on by policymakers and stakeholders.

The thalassemia prevention act is in the formulation stage, after the legislation, the policy rules will be set up for implementation. Effective implementation depends upon the strong basis of formulation that relies on the well-defined policy protocols. The well-formulated policy brings non-detrimental outcomes to fulfil the policy objectives. The legislators who participated in the study discussed the policy guidelines in detail. A legislator who was part of the legislation of previous health- related laws shared his views and said:

> The people involved in the thalassemia prevention act are not new to policy making and this is not the first bill that is going for presentation in the assembly. We will definitely follow the already present policy guidelines. The

> only thing needed for the execution is commitment. If the government is committed, it will happen in no time.

The study finds through this discussion that the lack of policy protocols and guidelines is not an obstacle to premarital screening policy formulation. The guidelines are already present in the form of previous policies and similar guidelines can be developed for newly formulated policies. The study participants also do not consider it a barrier to policy formulation.

Knowing the facts: Heading towards evidence-based policy execution

Evidence-based policymaking is an integral part of the policy process. It uses the approach to use already existing evidence for designing a policy and guiding future decisions. Evidence-based approach includes research, data collection, data collection management and using those research findings to execute policy and programs related to human services like health and education. Governments and agencies can benefit from an evidence-based policymaking approach. Some of the policy domains still lack the skills and capacity for effective use of evidence for integrating the policy formulation and implementation process. The research finding and evidence-based approach used in policymaking can benefit in many ways. It can inform policy decisions, create a knowledgeable environment, reduce uncertainty in policy decisions and eliminate the political divide. For this reason, policymakers have pressure to produce evidence-based policies and are subject to the justification of their policies. They are able to do so on the basis of research findings, empirical data and academic research data available (Leuz, 2018).

Evidence-informed policy decisions in public health are not only about using the research findings for the policy decisions, but it is to connect the political choices with the existing knowledge and evidence (Cairney & Oliver, 2017). The difficulty that the policy process faces in the way of legitimate policy execution is the absence of a combination between scientific evidence and political will or consideration. In the case of prevention of thalassemia through premarital screening, very less scientific research is present. A participant said in this regard, “Unfortunately, there is lack of scientific research in the area of prevention of genetic diseases. The evidence used by policy makers is not gained by scientific research.” Moreover, the research and political mood for policymaking have to be coordinated for a successful policy using the evidence-based decision-making approach. Almost all of the participants agreed on the need for evidence-based policymaking. A policymaker from the specialised health care department shared his views and commented:

> It is true that policies based on scientific research have stronger foundations and show positive desired outcomes but very limited research is done for premarital screening of genetic diseases and thalassemia. The research conducted is academic, and does not have much influence on policy. Moreover, policies are not based on evidence and they are often politically focused, policymakers in this scenario do not pay much attention to the inclusion of research in the formulation and design of the policy.

Limited research has been conducted on premarital screening of thalassemia in Pakistan. A few scientific and academic research is conducted but not focusing on policy formulation. The need for evidence-informed policy formulation is an important factor in policy decisions the policymakers and legislators take. The policies formulated after research will have firm grounds and effective outcomes.

Data organisation

Data organisation is also a part of evidence-based grounds of policymaking. When scientific research is done, data organisation is an integral part of compiling the evidence in the form of research findings. As discussed earlier, the policy decisions does not use scientific evidence for formulation and implementation, similarly, if some policy-focused research exists, the data is not organised in a way that it could be effectively used for the decision-making in the legislation of new laws and policies.

While discussing in detail the prevention of thalassemia, the present study explored whether there is a lack of research on genetic diseases and their prevention, research is not focused on policy making, or the scientific data is not organised so that it can be used for policy decisions. The study participants identified it as a major barrier in lack of research-based policy. A policymaker from the prevention department commented.

> Well… this is really a problem that there is very limited research done in the field of genetic diseases and specially premarital screening. Moreover, the bigger problem than this is the organisation of data. Even if the scientific evidence is present, the skilled data organisation lacks and eventually the idea of evidence based policy focus declines producing unsuccessful results of policy.

Efficient efforts for organising the data and research evidence are needed to manage the evidence-based policymaking skilfully. This may rely on the organisation of existing data in a skilful manner so that well-designed screening policy with strong evidence-based foundations emerge and maximum effectiveness of the formulated policies can be achieved.

Translating research into policy language: Interpretation of evidence

Like the organisation of data, the interpretation of the evidence is also a challenge in evidence-based approach of policy formulation. Interpretation and use of scientific evidence can help in policy suggestions and help the policymakers to understand the backdrop scenarios in a better way. Apollonio and Bero, (2016) suggested that interpreting the research evidence may not eliminate the political divide nor it can identify the problems or encourage the political will, but it is useful in suggesting policy alternatives. The evidence-informed policy decisions are desirable.

Premarital screening of thalassemia is an initiative to decrease the prevalence of thalassemia. The current study identifies that the research data that is under debate in the interviews with the study participants need to be clearly defined. A legislator said: “The interpretation of research data and perceiving the actual information it is intended for is very important”. There is a clear difference between the scientific data present that is intended to be used in the context of policymaking and the general information about the health problem. The policymakers commented on the interpretation of scientific research. A legislator who was the key informant of the Punjab Thalassemia Prevention Act expressed:

> Actually, we the stakeholders, including policymakers and legislators are generally confused about the concrete basis of the interpretation of evidence. The research is not designed and written in a way that it can be used for policy decisions.

Firstly, there is a lack of policy-focused research, which means that the scientific research is not done with a focus on policy making. Secondly, the present research and scientific evidence is either not understood to formulate policies or they are not interpreted in

a way they are intended for. This lack of interpretation of research data results in policies that are not evidence based and the actual picture of the problem often remains hidden. Policymakers need to involve the researchers in the process of policymaking so that they can interpret the research data according to the study intentions that can be used in the formulation of evidence-based policy.

Bureaucracy and policy making

Bureaucracy plays a vital role in bridging public needs with public policies made by governments. Bureaucrats are professional, trained and politically neutral members of the system. Bureaucracy links public needs with public policies. The major function of bureaucracy is to work for well-being and change through policy development (Srivastava, 2017). The present study explores the lack of collaboration between the bureaucracy and the government is a major policy barrier. Some policies result from the government's interest and bureaucracy is either overlooked or is so influenced that it interferes with the function of the bureaucratic administration.

Thalassemia prevention is a public health issue, which is much focused by the government and legislators, for this reason, the bureaucracy is not much involved in the policy formulation stages. The policymakers from the civil service talked about the collaboration of policymaking and bureaucracy. A bureaucrat said:

> The health minister is taking a personal interest in the prevention of thalassemia. This act has not faced any political opposition yet, but in the formulation phase, the bureaucracy is not much involved. I do not see this as a barrier to successful premarital screening law.

When political will is involved in policy making, the bureaucracy is not much involved in the process. Political will is the major factor needed for any task to be done. The bureaucracy however has a vital role in the dynamics of implementation of policy. The study participant further articulated:

> However, once the legislation has been done the bureaucracy will play a major role in implementing this law. Nevertheless, if the government will interfere in the administration with the pressure of other groups such as health care providers, this will definitely cause hindrances in successful implementation.

Political will and policymaking are correlated. Bureaucratic administration plays a vital part in the policymaking process. Collaboration between the policymakers and bureaucrats is an integral part of the policy discourse. There should be a partnership between the two, so that an effective policy in all the stages from policy design to implementation and outcomes is brought forward.

Discussion

Discussion

The present research explores the findings on barriers to the policy formulation and implementation of premarital thalassemia screening in Punjab and contextualizes them within global health policy literature. The results underscore how political, institutional, and socio-cultural factors collectively shape the feasibility of preventive genetic screening in low- and middle-income countries.

A key finding is the limited use of evidence-based policymaking. Respondents consistently indicated that inadequate research, poorly structured, and rarely integrated into decision-making. The findings that emphasis on the need of evidence based policy making, aligns with global challenges in translating scientific data into policy action (Cairney & Oliver, 2017; Apollonio & Bero, 2017). Moreover, this indicates the absence of robust national data on thalassemia prevalence, carrier rates, and cost-effectiveness, which hinders advocacy for mandatory screening. Leuz (2018) emphasizes that policies grounded in empirical data gain greater legitimacy and accountability. Therefore, strengthening local research capacity and data organization is essential to ensure evidence-informed policymaking and sustainable health legislation.

Political will, also emerged as a decisive factor in the study. The findings demonstrate that while the provincial government expresses interest, inconsistency in leadership priorities and administrative follow-up impedes progress. These observations are similar in the context of other health policies, such as HIV and cancer screening, where political commitment directly determines the success of implementation (DeMarco et al., 2011; Aljumah & Aljebreen, 2017). The study also suggests that participation of all the stakeholders is necessary for the success of policies. The interplay of all the components among policy making process is needed. Walt and Gilson's (1994) policy triangle model explains that actors, context, content, and process interact dynamically. Without political ownership, even well-formulated policies risk stagnation. The current study finds that sustained advocacy and bipartisan engagement are therefore necessary for the Thalassemia Prevention Act to move from the stage of draft to enforcement.

Another key finding identified by the study is 'bureaucratic participation'. Although bureaucracies are integral to translating policy into practice, their involvement during early policymaking stages remains minimal. Srivastava (2017) notes that citizen needs and policy outcomes are bridged by bureaucracies, and their exclusion undermines administrative efficiency. In Punjab, respondents observed that political leadership has largely dominated agenda setting, leaving bureaucrats to manage later stages. Institutionalized coordination between policymakers and civil servants could mitigate delays, improve communication, and strengthen accountability during policy execution.

Socio-cultural and ethical dimensions further complicate the procedure of implementation. Pakistan's high rate of consanguineous marriages reported at approximately 60% (Yaqoob et al., 1993) amplifies the genetic burden while posing cultural sensitivities. Religious reservations about abortion restrict antenatal options (Jafri et al., 2015), making premarital screening the most viable preventive approach. International experiences demonstrate success where screening programs incorporate religious and community endorsement. In Saudi Arabia and Iran, collaboration with clerics and public awareness campaigns significantly improved participation rates (Saffi & Howard, 2015; Al Sulaiman et al., 2008). Respondents similarly emphasized the inclusion of faith leaders and media to increase acceptance and counter misconceptions.

Economic analysis also reinforces the urgency of prevention. Treating thalassemia major patients through transfusions and iron chelation imposes enormous financial pressure costing millions annually (Ansari et al., 2011). For a resource-constrained health system, prevention is far more cost-effective. Yet, as Cornel and van El (2017) argue, developing nations often

prioritize communicable diseases and maternal health over genetic prevention. Demonstrating the long-term cost-benefit of screening through local pilot projects could help reposition thalassemia prevention on the national health agenda.

Public awareness and advocacy were recurring themes. Consistent with Alswaidi and O'Brien (2009), lack of knowledge and stigma surrounding carrier status remain critical barriers. Odelola et al. (2013) found that educational exposure increases positive attitudes toward genetic testing. Accordingly, integrating thalassemia awareness into secondary education, premarital counselling, and community outreach could enhance voluntary participation. Awareness efforts must also ensure confidentiality to protect participants from social discrimination.

Legal and institutional gaps were identified as another important challenge. Participants highlighted proposed drafts of the Thalassemia Prevention Act have been do not include detailed enforcement mechanisms or sanctions for noncompliance. Hussain and Ansari (2018) emphasize that the absence of legal frameworks and accountability weakens policy credibility. In contrast, countries enforcing mandatory premarital testing, such as Saudi Arabia, report consistent compliance when supported by legal safeguards (Al Sulaiman et al., 2008). It is recommended that future legislation on premarital screening of thalassemia should specify data protection protocols, counselling standards, and interdepartmental responsibilities to ensure ethical implementation.

The study also reveals broader implications for public health governance. Weak data systems, fragmented institutional linkages, and frequent political turnover mirror structural barriers common to other developing states. Policy research, data collection, and advocacy can be achieved by establishing a provincial genetics unit within the health department, that can coordinate with research and policy for a successful legislative procedure. Additionally, capacity-building programs for policymakers on evidence-based health legislation would bridge the gap between research and governance, ensuring more rational policy outcomes.

Conclusion:

The current research concludes that the success of premarital thalassemia screening in Punjab depends on multiple integral factors including political commitment, bureaucratic engagement, legal structure, research integration and social acceptance. Furthermore, evidence based, policy focused formulation awareness campaigns and dealing with challenges before hand are needed for effective policy. Prevention through screening offers both ethical and economic advantages, reducing the emotional and financial burden on affected families while safeguarding public health.

To transform this vision into practice, policymakers must adopt a holistic, evidence-driven approach that integrates scientific research with political and community collaboration. A barrier-free, well-coordinated policy process can enable Punjab and eventually Pakistan to progress from reactive disease management to proactive genetic prevention, setting a model for other low- and middle-income countries.

The study participants further supported the Government's notion of preventing thalassemia through premarital screening, as they are well aware of the fact that premarital screening will help reduce the burden of treatment on the health care system and the financial and emotional burden that families of thalassemia patients bear.

Authors' Contributions

Abida Tehreem: Conceptualization, Methodology, Formal Analysis, Investigation, Writing Original Draft, Visualisation, Writing-Review and Editing. ***Ahmed Usman***: Methodology, Formal Analysis, Supervision. ***Javeria Saleem:*** Conceptualisation, Validation, Supervision,

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