

A policy Analysis of Phenylketonuria Screening National Program in Iran

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Abstract

Background Phenylketonuria (PKU) Screening is a public health measure aimed at identifying the early cases and managing the affected infants and trying to reduce infant morbidity and mortality. The aim of this study was to analyze the policies of the PKU national screening program in Iran.

Methods PKU screening program policies were analyzed according to the policy triangle model. Document review and 38 semi-structured interviews were used for data collection. Document review data were analyzed using content analysis and interview data were analyzed using framework analysis.

Results The findings of the study were categorized according to the content (structure and organization, rules, bylaws and guidelines), process (agenda setting, formation, implementation, and evaluation of screening policies and programs), the policy context (political and executive factors, economic and financial factors, social and cultural factors), and the stakeholders (Genetics office, Network Development Bureau, Population and Family Health Bureau, Health Reference Laboratory, Pediatric Health Department, Health Vice-Chancellor of Medical Sciences, Health Ministry Deputy of Health, PKU Selected Hospitals, Pastor Institute, Food and Drug Administration and Research Centers, Welfare Organization, the Health Insurance Agency, the PKU Patient Support Association, the Post Company, and the Registry).

Conclusions The policy triangle framework helped to explain the different components of the PKU screening program.

Background

Phenylketonuria (PKU) is the most common hereditary metabolic disorder in the world (1). Patients with PKU show high levels of phenylalanine in the blood serum, and thus develop neurological disorders such as mental retardation, seizures, behavioral problems, and developmental delays (2–3). The prevalence of the disease is reported to be 1 in 10,000 in whites and 1 to 8,000–6,000 in the Iranian population (4). Other internal studies have also reported a high incidence of the disease among people with mental retardation from 1/2–5% (5–6). If the disease is not diagnosed and treated within the first year of birth, the child's IQ capacity can be reduced by 50%. Irreversible effects on the brain can be prevented by timely detection and control of phenylalanine levels in the blood (7).

Neonatal screening is a population-based public health screening program implemented for early detection (8). PKU screening is a prerequisite for early implementation of the Phe restricted diet, which is essential to prevent severe neurological disorders in patients with PKU (9). Nowadays, most developed countries carry out PKU screening, and any newborn with PKU is immediately exposed to a Phe-restricted diet to lower plasma Phe concentrations. This combination of early diagnosis and initiation of treatment results in normal IQ for most PKU patients (10).

The PKU screening program in Iran began in 2006, and blood samples were taken from all infants on day 3 to 5 after birth for colorimetric screening. And people with phenylalanine levels of 4 mg/dL or higher were referred to be confirmed by HPLC test. Then regular follow-up is performed for people who have phenylalanine levels equal to or greater than 4 mg/dL, and if the phenylalanine levels are 7 mg/dl or more, the diet begins with restricted phenylalanine. Dietary supplements of Iron, zinc, selenium, carnitine, vitamins and essential fatty acids are prescribed for all children up to 2 years of age (11).

Policy analysis includes plans and actions to achieve health care goals by defining a vision for the future, setting goals and objectives, setting priorities, and identifying the roles of different groups. Health policy analysis helps to understand the success or failure in implemented policies so that they can be useful for planning future policies. Health policy analysis helps policymakers to improve their successful implementation of future policies and provide opportunities to produce policy documents (12). Policy analysis can help to understand the complexity of the policy process and its nature and provide policy-related evidence (13). Policy analysis helps to understand why policymakers pay attention to certain issues in the health system and disregard other issues, also to identify stakeholders who agree or disagree with the policies and the reasons for or disagree with it, as well as to identify undesirable consequences of policies implementation and future problems in implementing policies and achieving their goals (14–15). Studying the role of evidence in policymaking helps to better understand the contribution of research to policy formation and to identify the factors that influence it. However, research evidence in policymaking is still less used in the eastern Mediterranean region (16). Therefore, this study aimed to analyze the policies of the national phenylketonuria screening program in Iran.

Methods

Study design

This qualitative study was conducted from May 2015 to January 2016 using the Walt and Gilson triangle framework. The model covers four general areas: content, context, actors, and the decision-making process. The content includes policy goals, operational policies, and so on. Actors refer to executives and influential organizations. The context refers to social, economic, political, cultural and other environmental conditions. The process consists of four parts: agenda setting, policy formulation, policy implementation, and policy evaluation (17).

Study population and selection of participants

Study participants included 38 individuals from the Health Commission of the Islamic Consultative Assembly, Ministry of Health (deputies of health and treatment, food and drug administration, the Iranians Health Insurance organization, the policymaking council, faculty members in universities of medical sciences, children's specialized hospitals, Pasteur Institute), Welfare Organization, and PKU Patients Support Association. Target groups were selected based on purposive sampling. Their selection criteria included knowledge and experience in phenylketonuria screening programs, active participation in phenylketonuria screening programs, and interest in research participation.

Data collection

Semi-structured interviews were used to collect data using the interview guideline. Credibility, transferability, dependability, and conformability were used such as criteria of rigor and trustworthiness of this study. Interviews were conducted face-to-face at a designated time and place. The interviews lasted between 30 and 65 minutes. At the beginning of each interview, a summary of the research topic and the method of using the data were provided for the participants. The written consent was obtained to conduct the interview. Noting was also used during the recording of the interviews. The audio files were transcribed at the earliest opportunity.

Data analysis

Framework Analysis was used to analyze the data, which included five basic steps: Familiarization, Identifying a Thematic Framework, Indexing, Charting and Mapping and Interpretation. In addition, Document Analysis was used to validate the findings of the interviews and to profit from the evidence available in the policy process. Documents were collected purposefully by referring to the organizations involved in the national phenylketonuria screening program and reviewing internet sites. The content analysis method was used for data analysis of documents.

Results

Context

Political and executive factors affecting program implementation include support for preventive programs in Iranian Development Plans laws, Patient request from Ministry of Health, relatively rapid changes in the Ministry of Health, job instability of team treatment at the hospitals and the shortage of manpower and equipment needed in the laboratories, working in parallel with the relevant scientific and executive organizations". The system should not change with the change of managers. It must be stable. After the management, specialized staff also changes. Then the problem happens "(P.19)."

The economic and financial factors affecting the implementation of the program included the effect of economic sanctions on drug and Phe-free milk imports for patients and the publication of scientific articles, inadequate allocation of funds and financial problems for families of patients.

"Iran's sanctions have made us constrained by drug access and low access to technology" (P.17). "Families pay most of the costs. This is not a disease that to treat it and say to the patient you can go and comeback next year. Intervals between following ups are short and this creates a financial burden for families "(P. 7).

Social and cultural factors affecting program implementation include positive attitudes towards the program and more reference in order to disease prevention, weak parenting skills of parents, poor self-care in adolescents, withdrawal from treatment in adulthood, and lack of internal control, the desire for family marriages in society and social stigma associated with the disease."With age, withdrawal from treatment increases and disease control becomes more difficult. Many patients develop seizures because they have not followed the diet and their blood phenylalanine levels have risen." (P.28). "Disease-related social stigma makes it difficult to care and follow up. The

parents of one of the patients changed their home so that they would not be followed up so that they would not be found. A person who has a child with PKU doesn't even tell his sister and brother that his child has PKU" (P.36)

Content

The purpose of phenylketonuria screening national program was to reduce physical disability, mental retardation and family damage. Phenylketonuria screening program was organized at the county, provincial, and national levels. The Genetics Department of the Ministry of Health was designated as responsible for implementing the program nationwide. In the health section, health deputy of medical universities and in the treatment section, children's specialized hospitals as PKU selected hospitals implemented the program in the provinces. In order to implement the program, guidelines of laboratory, care, clinical, nutritionist, genetic diagnosis and prevention, non-classic sampling guidelines and supplementary regulations of clinical psychologists were developed. In addition, a joint memorandum was signed between the Ministry of Health and the Post Office to transfer the laboratory samples. The infant's milk powder was subsidized by the Food and Drug Administration.

Policy formulation

Agenda setting

The problem stream can be attributed to physical and mental disabilities, IQ loss, and the costs of not being diagnosed early (First Stream). "There was a problem. Every year a number of patients were added to PKU patients. They suffer from severe physical and mental disabilities, reduced IQs and complications such as seizures and increased financial burden for the families and health system. On the other hand, the disease was able to be diagnosed early and be cared for.(P.1).

The above problems have led health policymakers to find a solution to these problems. Therefore, with the successful implementation of the hypothyroid screening program and the use of Infrastructure of this program for phenylketonuria screening, as well as executive and scientific efforts PKU screening program was developed (Second stream)."We had already experienced congenital hypothyroidism screening. Sampling time, sampling method and sending to the lab is the same as phenylketonuria screening".(P.14).

Despite opposition from the Network Development Office to integrate the program, insufficient support of insurance organizations and deputy of treatment of ministry of health, the Phenylketonuria national screening program was implemented due to the continuous persistence of parents and their demand to initiate screening and therapeutic interventions, interest, and perseverance of executives at the Genetics Bureau to convince policymakers. (Third stream (

We, patients' families, corresponded with the Vice- Chancellor of Health Minister. He met with us. We talked to him about our problems. He was very upset about our situation. He ordered all PKU patients to be served (P.22). One of the reasons for the success of this program was the presence of a capable, interested, and persistent responsible person. (P.16).

Formulation of phenylketonuria screening policies

The scientific proposal for the implementation of the screening program was formulated by two faculty members. A services package to patients with hereditary metabolic diseases was developed by faculty members from various universities. Program feasibility was performed by different health groups. The results of the health system evaluation for the implementation of the program showed that from the perspective of some officials, the PKU program was not a priority. Documentary statistics from PKU were not available at the start of the program. There was no complete information system to provide up-to-date information on the disease. There was a lack of HPLC to confirm the tests.

Implementation of PKU screening policies

The approach to implementing PKU screening policies was a top-down approach. The pilot program was conducted in 2007 at Universities of Medical Sciences Shiraz, Mazandaran and 3 Universities of Medical Sciences in Tehran. The screening program was nationwide after the pilot in early 2013. Organizing the laboratory tests, transferring the specimens and confirming the tests were done by the Health Reference Laboratory. In the center of each province, a children's specialized hospital was organized as PKU's selected hospital. A treatment team of pediatrician, nutritionist, psychologist, social worker, and secretary was formed in these hospitals. Patients' dietary milk was distributed by the selected hospital pharmacy. At the beginning of the program, a connector was selected from the parents of the patients, but after a while, this role was assigned to the hospital staff. The follow-up of the newborn baby is monitored by

health center experts. Detailed statistics of treatment and absence of treatment from the selected hospital were provided to the Ministry of health. Malignant and non-classic diagnostic tests were conducted at the Pasteur Institute.

Evaluation of PKU screening policies

Provincial experts' performance is evaluated by the Genetic Department of the Ministry of Health. In this way, periodic monitoring and completing checklists and providing statistics and documentation is done.

Challenges of Implementing a PKU Screening Program

A national screening decision was made in 2011, but was delayed due to laboratory equipment problems in medical universities in 2012. Monitoring and follow-up of PKU patients have lower priority than other programs in some selected hospitals. Selected hospitals suffered from a shortage of milk powder. It was not appropriate to record hospital and non-hospital information. Qualitative evaluation of selected hospitals was not performed well due to inadequate co-operation of some hospital managers and the lack of a supervisory system for providing PKU services in hospitals.

From the year 2007 to 2011, when we wanted to start a national screening program, during these four years, the most problems were laboratory diagnostic problems "(P3)."One of the problems is that hospital information is not recorded very well. (P2).

Actors

Internal stakeholders involved in the PKU national screening program include Genetics department, Health Network Development Center, Office of Population & Family Health, Health Reference Laboratory, Children Health Department, Deputies of Health Universities of Medical Sciences, Deputies of treatment Services, Selected Hospitals of PKU, Pasteur Institute, Food and Drug Administration and related research centers were external stakeholders involved in the Welfare Organization, Health Insurance Organization, PKU Patient Support Association, the Post office and Registration Office.

Discussion

The Phenylketonuria Screening National Program was announced because of pressure from parent persistence and their demand for screening and therapeutic interventions and interest, follow-up, and persuasion by executives to persuade policymakers.

The results of Barojo study in 2007 showed that policymakers' willingness and pressure of families with affected children led to a particular disease being selected for the neonatal screening program in Latin American countries without any appropriate basis or the use of a national standard (18). In the United States, legal pressure from parents and the legislature was the main driving force behind the widespread screening (19).

The faculty members of the medical sciences universities and accredited guidelines were used to design the program. New Zealand, which launched the Metabolic Disorders Screening Program since 2005, used a consulting group including pediatricians, patients' families and other medical teams before implementing the program (20). Phenylketonuria treatment team includes pediatric endocrinologist, nutritionist, psychologist, social worker and secretary. The results of the Hanley study in Canada showed that health care workers for the phenylketonuria screening program were physicians, nutritionists, nurses, social workers, biochemists, genetic counselors, and psychologists (21). Midwives, neonatal unit staff, health visitors and health team members, nursing specialist consultants, pediatric nurses in the pediatric nursing team, child health team, public health staff, general practitioners and pediatricians are involved in the implementation of the UK Neonatal Screening Program (22). The use of nurses and midwives in the care team of patients with phenylketonuria in Canada and the United Kingdom was a distinctive feature of the structure of human resources with Iran.

Based on the results, a "top-down" approach to policy implementation was adopted. This approach is focused on a small group of high-level policymakers and policies are seen as the orders of the authorities that the lower levels need to implement. In this approach, there is no chance for bottom-up negotiation and communication to influence the policy process (23).

The results of the study showed the impact of political changes in the country and consequently structural and managerial changes in the health system which have an adverse effect on the implementation of phenylketonuria national screening policies and programs. This result is in line with the results of the studies by Romain et al in 2015 in Tunisia (24). Unlike many developed countries in which the relocation of managers is not related to political change, in Iran, the position of executives and policymakers is weakened by any political event that stops programs (23).

Another political factor was the impact of international sanctions on imports of dietary milk and medicine. Studies by Masoumi et al in 2015 (25), Georgia et al in 2014 (26) and Shahabi et al in 2015 (27) have also addressed the impact of sanctions on health. The sanctions have had a negative impact on the country's treatment system, which includes the provision of drugs and medical equipment. In many cases, it has impeded the entry of certain drugs and has also caused slow entry of drugs and equipment, the introduction of counterfeit and inappropriate drugs, and increased prices for other drugs (28). One of the barriers to the implementation of the program was the cost of diagnostic, medical, and rehabilitation services for families, leading to increased out-of-pocket payments and lack of follow-up and timely treatment of patients. The results of Padilla & Terrell study in 2007 and May et al. (30) and Barajo study in 2007 (18) were the challenges of implementing the economic screening program for infants. The results of a 2003 study by Han and Lee in South Korea showed that limited government funding has led families to pay for phenylketonuria testing (31).

One of the important cultural factors was the family's lack of awareness of phenylketonuria. This result is consistent with the results of a study by Hong et al in southern Taiwan in 2005(32) and Barajo study (33), but it was inconsistent with the results of Campbell and Ross's study in the US in 2003(34). Parents' ignorance of the disease can be due to their poor health literacy.

Study Limitations

The study also had some limitations. It was difficult to study the participants' cooperation in the study, and few studies were conducted in this field.

Conclusion

The policy triangle framework helped to explain the different components of the PKU screening program.

Abbreviations

PKU
Phenylketonuria
IQ
Intelligence Quotient
HPLC
High performance liquid chromatography

Declarations

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Conflicts of interest

There are no conflicts of interest.

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