

**National registry of Haemoglobinopathies in Pakistan**Kamal Huda Khan<sup>1</sup>, Hania Zahid<sup>2</sup>, Abbas Mustafa Gain<sup>3</sup>, Naseha Mushtaq<sup>4</sup>, Rabeea Mushtaq<sup>5</sup>

Dear Editor, A national registry for hemoglobinopathies in Pakistan, a concept quite foreign to the country's healthcare system, has never been brought forth or implemented by its officials at a national level. When using the word registry and hemoglobinopathies we simply mean a centralised system where all the cases of haemoglobin-based disorders are registered and accounted for. Even though it may seem trivial, a centralised system can bring significant improvements to our healthcare system. A centralised data collection enables understanding disease burdens, guiding evidence-based policies, efficient resource allocation, early detection, and proper management. It supports research, genetic counselling, international collaboration, and connects patients with vital healthcare resources.

Worldwide data demonstrates implication of registries in various healthcare systems promoting disease scrutiny, detailed studies on health conditions and the improvement of management protocols with respect to the changing circumstances.<sup>1</sup> In Europe, the eENERCA project, helps in taking preventive measures against major rare anaemias (RAs) by facilitating diagnosis and therapeutic management.<sup>2</sup> In Saudi Arabia, Third Royal Decree in 2004 mandated premarital testing for sickle cell anaemia and Thalassemia, as consanguineous marriages are linked to the spread of hereditary diseases. Incorporation of this initiative assisted in reducing the rates of consanguineous marriages and, consequently, inherited diseases.<sup>3</sup> In Iran Since 1995, there has been a decline in Beta-thalassemia births up to 90.13% by implementation of the Thalassemia Prevention Programme. Similar to Iran, Greece and Cyprus were able to completely eradicate thalassemia despite having

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higher thalassemia burdens than Pakistan by adopting similar initiatives.<sup>4</sup>

Various studies in Pakistan were carried out to analyse the prevalence of haemoglobinopathies in different cities. A study carried out in Karachi through retrospective blood analyses revealed that the prevalence of haemoglobinopathies was 34.2%, comparable to a study conducted in Islamabad that revealed a frequency of 28.4%.<sup>5</sup> Also, in a different investigation that took place in northern Pakistan, out of 1187 refractory anaemia cases, 305 (25.69%) had thalassemia or abnormal Hb. In Pakistan through several studies, it has been suggested that Thalassemia is the most commonly occurring haemoglobinopathy.<sup>5</sup>

Considering the current state of increased rate of haemoglobinopathies in Pakistan, incentives need to be taken for ensure efficient control of the disease. Establishment of disease registries would promote execution of efficient disease preventive measures and organisations, thereby allowing implementation of effective management plans. Provision of adequate resources and funds by the government would help ensure high functioning registries. Commencement of awareness campaigns related to the knowledge of the disease and associated risk factors, screening programmes antenatal and premarital, and effective counselling techniques can help in reducing the incidence of haemoglobinopathies. Moreover, the registries will aim to build friendly relationships with other countries whereby innovative measures to boost the efficacy of registries will be suggested and implemented. On a patient level, the registries are believed to hold credibility in lowering the incidences of hereditary diseases and improving lifestyles of the affected.

**DOI:** <https://doi.org/10.47391/JPMA.22916>**Disclaimer:** None.**Conflict of Interest:** None.**Source of Funding:** None.

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### AUTHORS' CONTRIBUTIONS:

**KHK, HZ, AMG, NM & RM:** Concept, design, data acquisition, analysis, interpretation, drafting, revision, final approval and agreement to be accountable for all aspects of the work.