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Article

Haematologists as Genetic Counsellors for Haemoglobinopathies: Are They Prepared?

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Abstract

In haematology, a wide range of blood disorders are hereditary. The thalassaemias are hereditary anaemias characterised by a high burden of disease at the public health level, challenging the resources of many health systems. This review focuses on thalassaemia for which many countries have developed screening and prevention programmes. To manage this heavy burden two approaches were introduced include the reduction of new affected births aiming to keep the patient population from increasing continuously, and at the same time, offering parents a chance to informed choice concerning their reproductive life. Genetic counselling is a vital component of prevention aiming to inform individuals who are found to be carriers and couples who are both carriers in the case of recessive inheritance, of the possible consequences. These are all complex issues which may have to be discussed and challenge the skills which a genetic counsellor is expected to possess and utilise in every counselling session. The concern is that such trained and skilled scientists are few in numbers. It is obvious that for blood disorders counselling is rarely in the hands of qualified counsellors. Currently, in almost all haemoglobinopathy prevention programmes, counselling is offered by the clinicians in charge of clinical care. It is necessary to incorporate genetic counselling as an integral part of haematology services. The Thalassaemia International Federation suggests special training as part of haematology training, considering the complexity of the issues that must be discussed and a multidisciplinary approach to counselling should be considered where possible.

Keywords: hereditary anaemias; thalassaemia; genetic counselling

1. Introduction

In haematology, a wide range of blood disorders are hereditary. These include the haemoglobin disorders (haemoglobinopathies), bleeding disorders, and rare anaemias. This review focuses on thalassaemia for which many countries have developed screening and prevention programmes, although some aspects of screening and prevention could be widely applied to other haemoglobinopathies like sickle cell disease.

The thalassaemias are hereditary anaemias characterised by a high burden of disease both at the public health level, challenging the resources of many health systems, as well as at the patient and family level. This is attributed to the high prevalence in many populations across the world and their lifelong requirement for optimal care, including a constant supply of blood for red cell transfusions, expensive medications, and careful monitoring with multidisciplinary care. They are caused by mutations in the globin genes of the haemoglobin molecule, resulting in chronic anaemia due to ineffective erythropoiesis and haemolysis, iron overload, and organ damage. Even though severity varies, optimal care including clinical monitoring from infancy and throughout life is required to ensure longevity and reduction of comorbidity.

Haemoglobinopathies are the most common severe monogenic disorders globally, with 7% of the global population being carriers of a pathological haemoglobin gene and more than 500,000 affected children are born annually. Overall, the estimated prevalence of thalassaemia is higher in the Middle East, Asia, and Mediterranean countries than in Europe or North America, while

migrations are changing prevalence across these regions [1,2]. Sickle cell disease on the other hand has highest prevalence in Africa (~800/100,000), followed by the Middle East (~200/100,000), and India (~100/100,000), in contrast to ~30/100,000 in Europe with population movements carrying more affected individuals across continents [3].

In thalassaemia, chronic transfusions aiming to reduce ineffective erythropoiesis and subsequent anaemia lead to iron overload and damage to vital organs. This can be prevented by daily iron chelation therapy but imposes an additional burden on both health services and the individual patient. If not carefully applied, complication rates become high with monitoring and treatment requirements adding to the overall clinical burden of the disease [4,5].

In order to manage this heavy burden, apart from continually improving clinical care, two approaches were introduced. The first, practiced from the 1970s, is the reduction of new affected births aiming to keep the patient population from increasing continuously, and at the same time, offering parents a chance to informed choice concerning their reproductive life [6]. The second approach was to develop curative treatments and haematopoietic stem cell transplantation from HLA compatible donors has been available since the 1980s [7]. Scientific advances have also led to the development of genetic therapies such as gene addition or gene editing [8]. Along with these approaches, new pharmacological approaches are emerging to reduce the burden of anaemia and transfusion requirement, through medications such as luspatercept (promoting erythroid differentiation) [9] and mitapivat (managing red cell health by activating pyruvate kinase) [10], and others. However, such new developments increase costs and are unaffordable to patients living in developing economies, inevitably increasing inequalities in the global patient community.

All these issues are or should be known to treating physicians who are usually paediatric or adult haematologists (primary care providers, paediatricians. and internists may also be involved). Haematologists should also be heavily involved in national prevention programmes, since they have expertise in laboratory diagnosis as well as other aspects of prevention.

Prevention programmes are generally centrally controlled national programmes, which are based on raising public awareness, screening to identify asymptomatic carriers preferably before conception, and genetic counselling to be followed by possible interventions such as preimplantation testing and prenatal diagnosis, according to the at-risk individual's or couples' informed choice [5]. Such interventions are of vital importance to the wellbeing not only of the potential offspring, but also of the carrier couples at risk, the potential parents who may have to make painful decisions about their future and about their relationships.

We are aware of 25 countries where some effort at preventing or reducing thalassaemia births is ongoing. In a minority of countries, these are complete national policies including all facilities and offering choices such as prenatal diagnosis or pre-implantation genetic testing. In many others, the programme stops at screening for premarital couples or in antenatal clinics. In all cases, however, genetic counselling is or should be offered to individuals or couples at risk.

2. Counselling

Genetic counselling is a vital component of prevention aiming to inform individuals who are found to be carriers and couples who are both carriers in the case of recessive inheritance, of the possible consequences, either to their own health or to their offspring. In the case of haemoglobinopathies, the vast majority of detected carriers are asymptomatic and can be detected by haematological tests supplemented by molecular tests. If their partner is also a carrier, then in the recessive mode of inheritance, there is a one in four chance in each pregnancy of the child being affected by a syndrome which may be severe, or relatively mild according to the genes and genetic modifiers that are involved.

The choices available for carrier couples are listed in Table 1, which include major decisions with serious emotional and social consequences. Cultural differences, religious influences, legal considerations, literacy, as well as personal preferences will affect decisions of individuals. In addition, policy makers give emphasis or direct the goals of prevention according to the same influences of culture. For example, avoiding ‘incompatible marriage’ is the emphasis of some national programmes [12]. Consanguineous marriage is a preferred practice in many countries which may influence the birth incidence and the prevention programme [13]. Other cultures offer solutions like prenatal diagnosis and termination of affected pregnancies [14] and pre-implantation genetic testing [15].

Table 1. *Choices available for couples “at-risk”.* Source: Eleftheriou A, Angastiniotis M. The Global Thalassaemia Review 2024. Thalassaemia International Federation; 2024 [11].

Risk Identified	Choices
Before marriage or pregnancy	<ol style="list-style-type: none"> To avoid marrying another carrier To separate from a relationship that puts their future children “at-risk” To marry their chosen partner, with knowledge of the risk involved
After marriage or cohabitation	<ol style="list-style-type: none"> To proceed with a pregnancy, accepting the risk of possibly bearing an affected child To avoid having children (e.g., choosing adoption) To undergo prenatal diagnosis, choosing to either accept an affected child or pregnancy interruption To use pre-implantation genetic diagnosis, as an alternative to prenatal diagnosis, and so avoid pregnancy interruption
When already pregnant	<ol style="list-style-type: none"> To undergo prenatal diagnosis (if in early pregnancy) To accept any outcome with no further action To interrupt the current pregnancy with no further action

Genetic counselling is an essential process that is necessary, whatever the culture, to offer knowledge by which affected people can freely choose what to do. It is a service that aims at addressing each individual separately and/or the couple in the decision-making process about assessing the possibility an affected child. It is of major importance to be considered a priority in all programmes and the expertise required must be recognised [16–18].

3. Who Provides Counselling?

Ideally, such counselling is provided by trained medical professionals known as genetic counsellors [19,20]. These professionals are trained in medical genetics, diagnostics, as well as in the communication skills of providing information and supporting individuals and families who through genetic testing have become aware of what may be shocking information that has to be digested so that actions are decided. Such actions are listed in Table 1 and can have a serious impact on personal and social life.

Some of the issues which the counsellor needs to address include:

- **Consent for pre-symptomatic testing:** This follows information about the reasons for testing and is satisfied to a very variable extent by an ongoing public awareness campaign. This again depends on issues such as literacy and the effectiveness of measures to reach the public such as using the media, flyers, lectures, or school teaching.
- **Interpretation of laboratory results:** This means knowledge of the screening algorithms and the laboratory tests performed. Knowledge of the mutations involved and genotype/phenotype possibilities. Sometimes it requires teamwork to reach a correct diagnosis and so an individual

counsellor must ensure that he/she has the correct final diagnosis to provide advice; or else if there is uncertainty this should be expressed and explained.

- An explanation on what being a carrier (heterozygote) means to the individual's health and future prognosis.
- Interpretation of family and medical histories and the family pedigree.
- Screening type related issues:
 - a. Premarital screening: decisions about marriage and possible separation can be a painful dilemma for individuals or couples.
 - b. Antenatal clinic screening: where the woman is found to be a carrier with an ongoing pregnancy, she is faced with the task of bringing her partner for testing, and the possible need for prenatal diagnosis, and finally the question of termination of the pregnancy if the foetus is found to be affected.
 - c. Family members: testing of other family members, especially where consanguinity is common. Is there an issue of disclosure of the presence of asymptomatic carriers in unsuspecting relatives? Does the proband want them to know or do they want to be screened? The right not to know must also be respected.
 - d. Possibility of non-paternity may arise and sensitivity around disclosure.
- Assessment of the chance of disease occurrence or recurrence.
- Education about the natural history of the condition, inheritance pattern, the diagnosis, management, and prevention.
- The affected offspring may be transfusion-dependent with a severe syndrome but may also have a milder non-transfusion-dependent syndrome which later in life may bring complications and increased morbidity. This may increase the dilemma to parents especially if there is a degree of uncertainty.
- The implications of having an affected offspring that may affect employment prospects and health insurance of the parents in some settings.
- The individual must be made aware of new advances in treatment and whether current or future management may alter the prognosis. The prospect of future therapies may influence a couple's decision.

4. Who Provides Counselling?

This relies on several factors:

1. Communication skills: these include active listening, privacy, patience, empathy, and building trust.
2. Ethical principles must be pointed out and elaborated: confidentiality and the non-directive approach leading to informed choice. The psychosocial and family factors that can influence choices must be understood by the counsellor. The autonomy of the couple must be respected. Some couples faced with difficult choices suggest or request that the counsellor should decide or direct the decision which should be theirs ('what would you do doctor?'). The counsellor must support the couple in the reasoning and decision-making process, while avoiding expressing an opinion.
3. Disclosure of the carrier status to other members of the family may be important especially where consanguinity allows for detection of increased numbers of carriers. Privacy laws may prohibit disclosure of information and so the choice to disclose rests on the carrier and not the counsellor, unless consent is obtained.
4. Cultural considerations: being sensitive to the religious and culture influences that may affect a couple's choice. This requires knowledge of various cultures but also the ability to explore the couple's beliefs (listening skills). Avoiding being critical of prejudices is important. Such

considerations are frequent in multi-cultural societies and are increasing following the migration flows of recent years.

5. Language can be a major challenge since messages may not be clearly understood by either side. A translator may be needed but is best not be a relative, in order to maintain confidentiality and uninfluenced choices.
6. Repetition of the session may be necessary since the first announcement of genetic risk may be emotionally tense making complicated concepts difficult to engulf at the first interview.
7. Informed choices are made following information offered in a non-directive manner, and considering the risk assessment, as well as considering family goals, cultural, ethical, and religious values. The emotional attachment of a couple as well as family and social considerations should be factored, understood, and respected.

Discussing choices after full information (Table 1) should rely on the following principles:

- Questions about choosing a partner or of separation or not, are for the individual or couple to decide free of any influences.
- Prenatal diagnosis is a choice, but methods, risks, and complications must be discussed. This will lead to the issue of possible termination of pregnancy which again is a matter of free choice.
- Pre-implantation genetic diagnosis including risks, costs, and religious acceptance must be discussed. The fate of stored embryos must be considered.
- Intra-uterine treatment of hydrops can be offered explaining the postnatal dependence on blood transfusion.

These are all complex issues which may have to be discussed and challenge the skills which a genetic counsellor is expected to possess and utilise in every counselling session. The concern is that such trained and skilled scientists are few in numbers.

5. Availability of Skilled Counsellors

According to relatively recent literature, the number of certified genetic counsellors globally is approximately 7000, practicing in at least 28 countries in 2018 [21]. Even if this number may have doubled in the last 7 years, the number of qualified genetic counsellors is still inadequate to meet the demand. If the estimate of 500,000 births affected by haemoglobinopathies each year is a good approximation, then for these conditions alone there are 1 million individual parents who theoretically would need counselling every year. It is obvious that for blood disorders counselling is rarely in the hands of qualified counsellors. The issue that arises is the quality of counselling offered by non-geneticists or genetic counsellors [22]. Comparing the outcomes of counselling by qualified counsellors vs clinicians provide variable conclusions. Rowley et al in 1995 [23] used indicators such as (i) the proportion of β -thalassaemia births after counselling, (ii) the knowledge of both partners "at-risk" after counselling, and (iii) whether the individual counselled brought his/her partner to be tested. The two groups differed only in the case of bringing the partner to be tested. They concluded that clinical service providers, following brief training, can provide as pedagogically effective genetic counselling for hemoglobinopathy carriers detected prenatally as can professional genetic counsellors. Nurses were as effective as physicians. However, the conclusion was that brief training was necessary.

Other attempts to assess the results of genetic counselling have been made. Increasing understanding, enhancing coping and adaptation, facilitating informed choice, and mitigating health risks are some of the outcome measures quoted [24]. In addition, new developments in genome research, potential cures and additions to the pharmacological armamentarium, require that the counsellor is well versed in these developments [25]. In this respect the practicing haematologist may be in a better position to inform the 'customers'. Moreover, the increased attention to the impact of lifestyle, environment, and other influences to health in the context of 'precision medicine,' make the art and science of genetic counselling even more demanding of specialised knowledge and understanding of all factors that may influence wellbeing and health. A further complication is the

increasing cultural and linguistic mix in many populations due to migrations, which require the counsellor to be aware of cultural considerations as well as social determinants which can influence outcomes. The skills required for psychosocial support during counselling are an added necessity for quality counselling.

In recent years, the National Society of Genetic Counsellors (NSGC) of the USA, has proposed standards for the 'critical assessment of quality' and for the reporting of genetic counselling as a 'complex intervention', and for evaluating genetic counselling outcomes. The final checklist consisted of 23 items classified within 8 domains (not all applicable in specific services) [26]. The checklist is an indicator of the complex nature of genetic counselling, illustrated in Table 2.

This section may be divided by subheadings. It should provide a concise and precise description of the experimental results, their interpretation, as well as the experimental conclusions that can be drawn.

Table 2. Standards for the Reporting of Genetic Counselling Interventions in Research and Other Studies (GCIRS). Source: Hooker GW, Babu D, Myers MF, Zierhut H, McAllister M. Standards for the Reporting of Genetic Counseling Interventions in Research and Other Studies (GCIRS): an NSGC Task Force Report. *J Genet Couns.* 2017;26(3):355-360 [26].

Domain	Examples of the items to be considered
Indication for genetic counselling	1. The reason for genetic counselling 2. Whether the counselee is asymptomatic or symptomatic
Other components	2. Genetic testing before, after or at the time of counselling 3. Other clinician interactions
Intervention delivery	1. Delivery mode (telephone, in person, telemedicine, couple, single) 2. Physical setting (hospital, clinic, public, private)
Provider of counselling	1. Qualifications 2. Number of healthcare professional involved
Risk content and communication	1. Basis of risk assessment (family history, test results, personal history) 2. Type of risk information provided (frequencies, absolute /relative risk)
Educational content	1. Educational tools (booklets, flyers, web applications etc.) 2. Educational models or theories applied
Psychotherapeutic content	1. Goals (decision making, family communication, facilitate coping and adaptation) 2. Psychotherapeutic models or theories
Duration	1. Length of the genetic counselling interaction 2. Number of genetic counselling interactions (including follow up sessions)

6. The Haematologist as a Counsellor

Of particular interest to this paper is the 4th domain of the NSGC standards (Table 2) which discusses the provider and his/her qualifications. We are assuming that fully trained and qualified genetic counsellors are not available for the majority of haemoglobinopathy programmes across the globe. Several universities outside the USA and Europe (including India with a huge haemoglobinopathy population) are offering courses in genetic counselling, and so in years to come many more qualified counsellors will be available. Currently, in almost all haemoglobinopathy prevention programmes, counselling is offered by the clinicians in charge of clinical care, laboratory scientists responsible for screening, and nurses. In some situations, antenatal screening of pregnant women is practiced and so the obstetric team act as counsellors.

In these scenarios and in view of the above-described requirements, is the quality of counselling assured? There is doubt whether these healthcare professionals are trained or whether they rely on their understanding of mendelian inheritance alone to provide risk assessment, without understanding the complexity of counselling. Yet, genetic counselling is an integral part of both patient care and the prevention programmes which are basic to haematology. Is a haematologist ready to discuss the impact of cousin marriage to an at-risk couple for example? The haematologist may be aware of the various mutations on the relevant genes but the phenotype resulting from various combinations must also be known before facing the couple. The haematologist may be the most expert to describe the various curative approaches but must also be ready to describe the advantages and disadvantages of each and avoid overoptimism which may affect a couple's choices. Emotional support is needed but it takes time to listen to concerns, time which clinicians may not have. Communication skills are variable among clinicians but are fundamental in any counselling session. Support, especially when uncertainty is a factor, can be a major challenge since it can increase patient anxiety.

7. Conclusions

This manuscript is not meant to convey that haematologists are not competent to offer genetic counselling. However, for most, prior training is necessary, and this has not been put forward in the past or even recent literature, as a requirement for qualification in the field. In view of the complexity of the service, another possible solution is that genetic counselling of these disorders is provided by more than one practitioner: either a qualified counsellor in collaboration with a clinician or a clinician with a psychologist/social worker or even with a laboratory scientist. Such combinations will depend on the skills and knowledge of the various members of the team. They may work together so that common decisions may be reached or have separate interviews with a final session to discuss the individual's or couple's impressions and decisions.

It is necessary to incorporate genetic counselling as an integral part of haematology services and one or more members of this specialty should acquire the knowledge and skills required for comprehensive counselling. Who provides training and for how long is an organisational question, although collaboration with a clinical genetics service or academic department is an important step.

To provide answers to the many questions that this topic gives rise to, research is needed. The difficulty is that research is most often carried out in countries where post graduate genetic counselling practitioners are most present and referred to in haematological counselling.

Final suggestions of the Thalassaemia International Federation are:

Genetic counselling is a complex service that the haematologist is called upon to fulfil as part of his/her duties.

Special training is recommended as part of haematology training, considering the complexity of the issues that must be discussed.

The possibility of a multidisciplinary approach to counselling can be considered where possible.

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