Global epidemiology of haemoglobin disorders and derived service indicators

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Abstract To demonstrate a method for using genetic epidemiological data to assess the needs for equitable and cost-effective services for the treatment and prevention of haemoglobin disorders. We obtained data on demographics and prevalence of gene variants responsible for haemoglobin disorders from online databases, reference resources, and published articles. A global epidemiological database for haemoglobin disorders by country was established, including five practical service indicators to express the needs for care (indicator 1) and prevention (indicators 2–5).

Haemoglobin disorders present a significant health problem in 71% of 229 countries, and these 71% of countries include 89% of all births worldwide. Over 330 000 affected infants are born annually (83% sickle cell disorders, 17% thalassaemias). Haemoglobin disorders account for about 3.4% of deaths in children less than 5 years of age. Globally, around 7% of pregnant women carry β or α zero thalassaemia, or haemoglobin S, C, D Punjab or E, and over 1% of couples are at risk. Carriers and at-risk couples should be informed of their risk and the options for reducing it. Screening for haemoglobin disorders should form part of basic health services in most countries.

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Une traduction en français de ce résumé figure à la fin de l'article. *Al final del artículo se facilita una traducción al español.* الترجمة العربية لهذه الخلاصة في نهاية النص الكامل لهذه المقالة.

Introduction

Inherited haemoglobin disorders (sickle-cell disorders and thalassaemias) were originally characteristic of the tropics and subtropics but are now common worldwide due to migration.[1–4] Since they can be controlled cost-effectively by programmes that integrate treatment with carrier detection and genetic counselling, WHO has recommended global development of these services.[5,6] However, service development can be unexpectedly challenging, because it requires inclusion of genetic approaches in health systems.

The diversity and heterogeneous distribution of haemoglobin disorders make it necessary to develop strategies at the country level. To assist policy-makers, we use haemoglobin disorders as an example to show how genetic epidemiological data can be interpreted in terms of administrative boundaries (and/or ethnic group) and practical service indicators. The work was initiated for WHO[7,8] and further developed in the United Kingdom,[9,10] where it is used for local needs-assessment.[11] Global data are available at: www.chime.ucl.ac.uk/work-areas/cab/.

Genetic terminology can be impenetrable to non-specialists. In this paper, we use terms proposed for more general use by the Professional Education for Genetic Assessment and Screening (PEGASUS) Genetic Education Programme of the United Kingdom National Screening Committee:[11] haemoglobin disorders include sickle-cell disorders and thalassaemias; haemoglobin gene variants are haemoglobinopathies; significant variants are gene variants that can cause a serious disorder; carriers are healthy heterozygotes; people with combinations of gene variants are homozygotes and compound heterozygotes; and α thalassaemia major is haemoglobin Barts hydrops fetalis.

Genetic background

Haemoglobin comprises four globin chains: fetal haemoglobin (Hb F) has two α and two gamma chains ($\alpha_2\gamma_2$) and adult haemoglobin (Hb A) has two α and two β chains ($\alpha_2\beta_2$). Genes in the α-globin and β-globin gene clusters (on chromosomes 16 and 11) control globin-chain production. Due to spontaneous mutation, haemoglobin gene variants are present at low prevalence (carriers 1–1.5/1000) in all sizeable populations.[2,12] They fall into two broad groups – structural variants that change the amino acid sequence and produce an unusual haemoglobin,[13] and thalassaemias that lower or abolish production of globin chains.[14] Most haemoglobin gene variants are rare and many are harmless, but some are common because carriers are less likely than others to die from falciparum malaria. The most common such variant, α plus (α^+) thalassaemia, is usually harmless.

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However, people who inherit combinations of haemoglobins S, C, E, D Punjab, β thalassaemia, or α zero (α^0) thalassaemia may have a serious haemoglobin disorder. In populations in which malaria is (or was) endemic, 3 to 40% of individuals carry one of these significant variants, and the prevalence of haemoglobin disorders ranges from 0.3 to 25 per 1000 live births.[8]

Carriers are easily detected by routine haematological methods and can be forewarned of their reproductive risk. Carriers of structural variants have 30–50% of the variant haemoglobin in their red cells: thalassaemia carriers have small red blood cells and sometimes mild anaemia,[3] and β thalassaemia carriers also have over 3.5% of Hb A_2. The resemblance between thalassaemia and iron deficiency can confuse the diagnosis of either disorder.[15]

Requirements for treatment

β thalassaemia major causes profound anaemia that kills untreated affected children before the age of 3 years. However, the life expectancy of patients treated with regular blood transfusion and iron-chelation therapy, or bone-marrow transplantation, is approaching normal.[16,17] α thalassaemia major causes hydrops fetalis and perinatal death, often with life-threatening obstetric complications for the mother,[3] and prenatal diagnosis usually leads to termination of pregnancy. Some cases have recently been saved by intrauterine transfusion, despite a high risk of severe mental and physical handicap.[18]

In sickle-cell disorders, sickled red blood cells block small blood vessels and cause anaemia, functional asplenia, episodes of severe pain, and residual organ damage. Most untreated affected children die from infection in early life,[19] but simple steps including neonatal diagnosis, prophylactic antimalarials and antibiotics, access to hospital treatment when needed, and information and support for families greatly improve quality and length of life.[20]

Requirements for prevention

A policy of detecting carriers and informing them of their risk, and possibilities for reducing it, usually leads to a fall in births and deaths of affected children. Requirements are the same for thalassaemias and sickle-cell disorders. In most countries, the approach develops in three stages.[21]

First, retrospectively informing parents with affected children of their 25% recurrence risk allows them to limit family size[22] and, where average family sizes are typically large, this approach can significantly reduce affected birth prevalence. Second, introduction of prenatal diagnosis for couples with affected children enables them to have a family, but has little further effect on affected birth prevalence.[21] Access may also be limited by economic, medical, social or legal factors. Third, information and prospective carrier screening is provided for the whole population. Choice of strategy varies with social attitudes, costs and opportunities within the health system. The offer of testing in high school[23,24] or before marriage[25–28] allows a wide range of choices and requires the least number of laboratory tests.[26,28] Screening during pregnancy[29,30] enables fewer options, requires more tests, is ethical only if prenatal diagnosis is freely available, and often identifies risk too late for the option of prenatal diagnosis.[31] When carrier screening is provided without the option of prenatal diagnosis it usually creates public demand for this service.[26,32] Population screening is not the only useful strategy: family studies can be cost-effective where consanguineous marriage is common[33] or carrier prevalence is low.[34]

The effects of screening depend on the choices made by informed individuals. Birth prevalence of thalassaemia can fall by over 90%[25–28,35] because most at-risk couples limit their family to two healthy children,[22,25,26] there is very high uptake of prenatal diagnosis, and some carriers avoid risk by selecting a non-carrier partner.[25,26] Available data for sickle-cell disorders shows lower use of prenatal diagnosis[36,37] and improved survival of affected children with neonatal diagnosis.

Methods

Acquisition of data

The necessary data sets are available for most countries. We gathered demographic data: population number, age distribution, crude birth rate and infant mortality from the 2003 United Nations Demographic Yearbook;[38] under-5 mortality from the United Nations Children's Fund (UNICEF);[39] supplementary information from national statistics on the internet, and the *Encyclopaedia Britannica*. Livingstone's 1985 database of 1351 epidemiological studies of haemoglobin disorders[2] provides robust global data on carrier prevalence (and so gene frequencies) for structural variants, but is less informative for thalassaemias. Data for thalassaemias were obtained from research reviews,[3,4] country visits, and the former WHO Working Group on Haemoglobin Disorders.[7] Data for α^+ thalassaemias are from Weatherall and Clegg.[3] Detailed sources and references are available at: www.chime.ucl.ac.uk/work-areas/cab/. For populations where consanguineous marriage is common, a population coefficient of consanguinity (F) must be included when calculating the prevalence of affected conceptions from gene frequencies.[40] Values for population F were obtained from Dr Alan Bittles' database[41] and older ethnographic sources.[42]

Calculation of birth prevalences and service indicators

Prevalences of conceptions with 12 combinations of gene variants were calculated for each country from gene frequencies using the Hardy–Weinberg equation:[40]

$$(p^2 + Fpq) + 2(pq - Fpq) + (q^2 + Fpq) = 1$$

where p is the gene frequency of variant 1, q is the gene frequency of variant 2, and F is the population coefficient of consanguinity.

The results are aggregated here into conceptions per 1000 of: (1) sickle cell disorders (SS, SC, S/β thalassaemia), (2) β thalassaemias (homozygous β thalassaemia, Hb E/β thalassaemia), (3) α thalassaemias (homozygous α^0 thalassaemia, α^0/α^+ thalassaemia), and (4) harmless combinations (CC, C/β thalassaemia, EE, DD, D/β thalassaemia, etc.).

The following five service indicators were obtained for every country by combining prevalences of carriers and affected births with demographic data.

1. **Indicator for patient care** (N) is the annual conceptions with a haemoglobin disorder in the absence of prevention. Where treatment is not available, N is a measure of childhood mortality due to haemoglo-

Table 1. **Estimated prevalences of carriers of haemoglobin gene variants and affected conceptions**

WHO region	Demography 2003				% of the population carrying			Affected conceptions (per 1000)			Affected births (% of under-5 mortality)
	Population (millions)	Crude birth rate	Annual births (1000s)	Under-5 mortality rate	Significant variant[a]	α^+ thalassaemia[b]	Any variant[c]	Sickle-cell disorders[d]	Thalassaemias[e]	Total	
African	586	39.0	22 895	168	18.2	41.2	44.4	10.68	0.07	10.74	6.4
American	853	19.5	16 609	27	3.0	4.8	7.5	0.49	0.06	0.54	2.0
Eastern Mediterranean	573	29.3	16 798	108	4.4	19.0	21.7	0.84	0.70	1.54	1.4
European	879	11.9	10 459	25	1.1	2.3	3.3	0.07	0.13	0.20	0.8
South-east Asian	1 564	24.4	38 139	83	6.6	44.6	45.5	0.68	0.66	1.34	1.6
Western Pacific	1 761	13.6	23 914	38	3.2	10.3	13.2	0.00	0.76	0.76	2.0
World	**6 217**	**20.7**	**128 814**	**81**	**5.2**	**20.7**	**24.0**	**2.28**	**0.46**	**2.73**	**3.4**

[a] Significant variants include Hb S, Hb C, Hb E, Hb D etc. β thalassaemia, α^0 thalassaemia.
[b] α^+ thalassaemia includes heterozygous and homozygous α^+ thalassaemia.
[c] Allows for (1) coincidence of α and β variants, and (2) harmless combinations of β variants.
[d] Sickle-cell disorders include SS, SC, S/β thalassaemia.
[e] Thalassaemias include homozygous β thalassaemia, haemoglobin E/β thalassaemia, homozygous α^0 thalassaemia, α^0/α^+ thalassaemia (haemoglobin H disease).

bin disorders. Where treatment is available, *N* indicates the potential annual increase in patients needing care, and enables cost projections.[43] Where prevention is available, *N* provides a baseline for measuring its effect on patient numbers.

2. **Indicator for carrier screening** is the annual carrier tests required. With antenatal screening this is the annual number of pregnancies (~ births) in risk groups. With premarital or prepregnancy screening, this is the annual number of young people in risk groups reaching reproductive age.
3. **Indicator for carrier information and offer of partner testing** is the annual carriers detectable by the chosen strategy.
4. **Indicator for expert risk assessment and genetic counselling** is the annual pregnancies to carrier couples, or new carrier couples, detectable by the chosen strategy.
5. **Indicator for the offer of prenatal diagnosis** is the annual pregnancies actually at risk (~ 3*N* to 4 *N*).

The indicator for neonatal screening for sickle-cell disorders differs with policy. When there is no adult carrier screening, all newborns in risk groups must be tested (indicator 2). When there is prior carrier screening, only infants born to carrier mothers (indicator 3), or to at-risk couples (indicator 5) may need to be tested.

Country estimates

The calculations use the most detailed country data available. These data range from limited historical surveys to detailed micromapping by geographical area or ethnicity. Estimates for 24 countries (including China and India) were derived by aggregating more detailed data. Estimates for 19 countries where haemoglobin disorders occur primarily as a result of migration were obtained by combining data on residents' ethnicity or country of birth with gene frequencies in countries of origin.[12] All estimates are the most conservative permitted by the data (i.e. give minimum figures). Individual country estimates are available at: www.chime.ucl.ac.uk/work-areas/cab.

Findings

Haemoglobin disorders were originally endemic in 60% of 229 countries, potentially affecting 75% of births, but are now sufficiently common in 71% of countries among 89% of births (either in the whole population or among minorities) to require policy-makers to consider the most appropriate strategy for treatment and prevention. Table 1 shows conservative prevalence estimates by WHO region. At least 5.2% of the world population (and over 7% of pregnant women) carry a significant variant. Haemoglobin S accounts for 40% of carriers but causes over 80% of disorders because of localized very high carrier prevalence: around 85% of sickle-cell disorders, and over 70% of all affected births occur in Africa. In addition, at least 20% of the world population carry α^+ thalassaemia.

Around 1.1% of couples worldwide are at risk for having children with a haemoglobin disorder and 2.7 per 1000 conceptions are affected. Prevention is making only a small impression: affected birth prevalence is estimated at 2.55 per 1000. Most affected children born in high-income countries survive with a chronic disorder, while most born in low-income countries die before the age of 5 years: haemoglobin disorders contribute the equivalent of 3.4% of mortality in children aged under 5 years worldwide or 6.4% in Africa.

Table 2. **Indicators of annual service needs for haemoglobin disorders**

WHO and component regions	Indicator 1				Indicator 2	Indicator 3	Indicators 4 and 5	
	Annual affected conceptions				Total annual births (1000s)	Annual pregnant carriers (1000s)	Annual pregnancies	
	Sickle cell disorders	β thalassaemias	α thalassaemias	Total disorders			Both parents carriers	At risk
African region	**233 289**	**1 520**	**11**	**234 819**	**22 895**	**4 363**	**1 005 752**	**939 277**
Northern Africa	181	337	0	518	627	35	2 882	2 073
Western Africa	167 224	971	0	168 195	9 622	2 551	738 373	672 781
Middle Africa	40 688	27	0	40 715	4 184	804	162 934	162 861
Eastern Africa	25 184	183	11	25 377	6 974	966	101 510	101 509
Southern Africa	11	2	0	13	1 487	7	53	53
American region	**9 047**	**533**	**442**	**10 022**	**16 483**	**523**	**44 769**	**40 088**
Northern America	2 637	268	429	3 334	4 435	122	15 780	13 337
Central America	175	2	0	176	3 627	20	724	705
Caribbean	3 333	16	0	3 349	778	4 505	14 369	13 394
South America	2 902	248	13	3 163	7 643	278	13 895	12 651
Eastern Mediterranean region	**6 491**	**9 715**	**1**	**16 207**	**16 776**	**670**	**66 079**	**64 828**
Northern Africa	1 456	1 829	0	3 285	3 776	152	13 986	13 140
Eastern Africa	8	19	0	27	3 067	19	109	109
Western Asia	4 479	1 815	1	6 294	3 540	218	25 178	25 178
South central Asia	547	6 053	0	6 600	6 393	281	26 806	26 401
European region	**1 292**	**1 347**	**162**	**2 800**	**10 459**	**153**	**12 064**	**11 201**
Northern Europe	429	82	73	533	1 162	15.0	2 660	2 333
Western Europe	387	78	56	521	1 949	23.2	2 556	2 085
Southern Europe	204	313	33	550	1 458	39	2 263	2 202
Eastern Europe	0	25	0	25	2 881	8	98	98
South central Asia	0	365	0	365	1 190	27	1 461	1 461
Western Asia	272	484	0	756	1 819	41	3 027	3 023
South-east Asian region	**26 037**	**21 693**	**1 383**	**49 114**	**38 139**	**2 363**	**421 398**	**196 454**
South central Asia	26 037	9 348	0	35 386	31 210	1 476	230 905	141 542
South-eastern Asia	0	12 345	1 383	13 728	6 929	887	190 493	54 912
Western Pacific region	**13**	**7 601**	**10 524**	**18 138**	**23 914**	**1 038**	**191 045**	**72 554**
Eastern Asia	0	1 672	8 106	9 778	18 592	420	39 129	39 110
South-eastern Asia	0	5 846	2 373	8 219	4 774	607	151 173	32 878
Melanesia, Micronesia, Polynesia	0	54	0	54	242	6	214	214
Australia and New Zealand	13	29	45	88	307	4.7	529	351
World	**276 168**	**42 409**	**13 466**	**332 043**	**128 667**	**9 111**	**1 744 877**	**1 328 172**

Table 2 presents the five service indicators by WHO region and geographical subregion. It also shows the rapid recent spread of haemoglobin disorders with migration (e.g. affected conceptions are now more common in northern and western than in southern Europe).

Indicator 1. Annually there are over 332 000 affected conceptions or births. About 275 000 have a sickle-cell disorder, and need early diagnosis and prophylaxis. About 56 000 have a major thalassaemia, including at least 30 000 who need regular transfusions to survive and 5500 who die perinatally due to α thalassaemia major.

Indicator 2. Most births, 75%, are in countries where haemoglobin disorders are endemic and 13% occur where they are common because of migration, so in principle, 88% of the 128 million women who become pregnant annually should be offered screening.

Indicator 3. Over 9 million carriers become pregnant annually. The risk that their partner is also a carrier ranges from 0.1–40% (global average 14%). In principle, all need information and the offer of partner testing.

Indicator 4. Annually there are at least 948 000 new carrier couples, and over 1.7 million pregnancies to carrier

Table 3. **Estimated reach of treatment for β thalassaemia in each WHO region**[a]

WHO region	Estimated annual births β thalassaemias		Transfusion			No. of known patients	Adequate iron chelation		Inadequate or no iron chelation	
	Total	Transfusion-dependent	Annual no. starting transfusion	% of transfusion-dependent patients transfused	Annual deaths because not transfused		% with chelation	No. with chelation	No. of patients	Annual deaths due to iron overload
African	1 386	1 278	35	2.7	1 243	–	–	–	–	–
American	341	255	134	52.4	121	2 750	58	1 604	1 146	57
Eastern Mediterranean	9 914	9 053	1 610	17.8	7 443	39 700	27	10 818	28 882	1 444
European	1 019	920	140	15.5	780	16 230	91	14 754	1 476	74
South-east Asian	20 420	9 983	962	9.6	9 021	35 500	19	6 621	28 879	1 444
Western Pacific	7 538	4 022	108	2.7	3 914	3 450	44	1 504	1 946	97
World	**40 618**	**25 511**	**2 989**	**11.7**	**22 522**	**97 630**	**39**	**37 866**	**59 764**	**2 988**

[a] All figures are minimum estimates.

couples. Around 75% are actually at risk. In principle, all need expert risk assessment and genetic counselling.

Indicator 5. Annually there are 1.33 million at-risk pregnancies. In principle, all need the offer of prenatal diagnosis.

Table 3 shows that about 12% of children born with transfusion-dependent β thalassaemia are actually transfused, and less than 40% of those transfused obtain adequate iron-chelation therapy. About 100 000 patients are currently living with regular transfusions, and at least 3000 die annually in their teens or early 20s from uncontrolled iron overload. No comparable data are available for sickle-cell disorders.

Estimated reach of prevention

Systematic carrier screening with the option of prenatal diagnosis is established in parts of Asia (in parts of China, including Hong Kong Special Administrative Region (SAR), Macao SAR, some southern regions and the province of Taiwan, parts of India, the Islamic Republic of Iran, the Maldives and Singapore), parts of the Caribbean and most of southern Europe (except Albania). In Australia, much of north-west Europe, New Zealand and North America, prenatal diagnosis is available and antenatal carrier screening is standard practice. In the United Kingdom this policy identifies only a minority of at-risk couples in time for a truly informed choice: for timely risk detection, screening must be provided through primary health care.[35] The same may apply for many countries where the disorders affect primarily ethnic minorities. The aggregated global data suggest a 16% reduction in births of children with thalassaemia and a 4% reduction in births of children with sickle-cell disorders. The greater part of the estimated reduction is attributed to reduced reproduction by informed at-risk couples, rather than prenatal diagnosis.

Discussion

Global burden of haemoglobin disorders

The yardstick of under-5 mortality can be used to assess the broad effect of haemoglobin disorders on health, because most affected children die in early childhood and most survivors have chronic disease. Table 1 shows that they cause the equivalent of at least 3.4% of deaths in children aged under 5 years. However, this still underestimates their burden because inherited disorders affect families. Worldwide, over 1% of couples are at risk for haemoglobin disorders, most have at least one affected child, and most affected children die in early childhood. Although the west African death rate in children aged under 5 years is 18.4%, the rate is 16.5% for children born to couples not at risk for sickle-cell disorders compared with 40% for children born to couples who are at risk. Clearly, methods to assess the health burden of inherited disorders must include a family perspective.[44]

Thalassaemias

Most children with thalassaemia are born in low-income countries. Worldwide, transfusion is available for a small fraction of those who need it, and most transfused patients will die from iron overload unless an available and potentially inexpensive oral iron chelator is licensed more widely.[45] The patients' predicament underlines the need for combined treatment and prevention programmes.[43] Wherever combined programmes exist survival is steadily improving,[16,17] affected births are falling,[25–28,30] and numbers of patients are stabilizing. The policy is spreading because of its demonstrable cost-effectiveness, and thalassaemia is gradually becoming contained.[43]

Sickle-cell disorders

In high-income countries that provide neonatal diagnosis and care for patients, most survive well into adult life[46] and, because there is limited use of prenatal diagnosis,[36,37] numbers of patients are

rising steadily. Most affected children born in low-income countries still die undiagnosed, usually from malaria,[19] but things are changing. About 40% of Africa is now urbanised, and improved access to health care is leading to increased survival and rising demand for hospital services.[47] Community-based services including information, prophylactic antimalarials or antibiotics, and social support greatly improve survival and quality of life and reduce demand for acute hospital services – in short, it is less costly to make organized care available than not.[47] If average survival reaches only half the African norm, over six million Africans will be living with a sickle cell disorder – clearly, care for these disorders must become part of primary care wherever they are common.

There is a strong case for carrier screening in Africa. Cheap and simple methods for testing adults and newborns exist. Knowledge of risk allows a range of options, including limiting of family size, ensuring that at-risk infants are tested at birth, and requesting prenatal diagnosis. DNA-based early prenatal diagnosis is available at several African centres and is relatively inexpensive when only the sickle variant is sought. However, as few couples can afford even a subsidised fee,[32] there is insufficient information on likely uptake if the service were freely available.

Relevance to diagnosis of iron deficiency

WHO recommends the use of haemoglobin concentrations to assess prevalence of iron deficiency in a lower-income setting.[48] However, the recommended cut-off values for haemoglobin concentrations are derived from populations of northern European origin and can lead to overestimation of iron deficiency where thalassaemias are common.[49,50] The high global prevalence of thalassaemias (Table 3) means that each population should use their own baseline normal ranges in the assessment of iron deficiency.

Conclusion

The data summarized here confirm that screening and genetic counselling for haemoglobin disorders should be an intrinsic part of health care in most countries, as recommended by the WHO.[5,6] The country estimates (available at: www.chime.ucl.ac.uk/work-areas/cab) provide a starting point for local needs assessment, service planning and evaluation. Because haemoglobin disorders are commonly a point of entry for genetic approaches into health systems,[43,44] services should be designed to provide a foundation for more comprehensive community genetics services.[26] ■

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Résumé

Epidémiologie mondiale des troubles de l'hémoglobine et indicateurs de service dérivés

Présentation d'une méthode utilisant des données d'épidémiologie génétique pour évaluer les besoins en matière de services équitables et d'un bon rapport coût/efficacité pour le traitement et la prévention des troubles de l'hémoglobine. Des données démographiques et de prévalence concernant les variants génétiques responsables des troubles de l'hémoglobine ont été réunies à partir de bases de données en ligne, de sources de références et d'articles publiés. Une base de données épidémiologiques mondiale sur les troubles de l'hémoglobine par pays a été mise en place, en même temps que cinq indicateurs de service pratiques, destinés à évaluer les besoins en termes de soins (indicateur 1) et de prévention (indicateurs 2 à 5).

Les pays où les troubles de l'hémoglobine sont un problème de santé important représentent 71 % des 229 pays considérés et totalisent aussi 89 % de la natalité mondiale. Plus de 330 000 nourrissons naissent chaque année avec de tels troubles (83 % avec une drépanocytose, 17 % avec une thalassémie). Les troubles de l'hémoglobine sont responsables d'environ 3,4 % des décès chez les moins de 5 ans. A l'échelle mondiale, 7 % environ des femmes enceintes sont porteuses d'une bêta-thalassémie ou d'une alpha-zéro-thalassémie, ou encore d'une hémoglobine S, C, D-Punjab ou E, et plus de 1 % des couples sont à risque. Les porteurs et les couples à risques doivent être informés des dangers qu'ils encourent et des solutions pour les réduire. Le dépistage des troubles de l'hémoglobine doit faire partie des services sanitaires de base de la plupart des pays.

Resumen

Epidemiología mundial de las hemoglobinopatías e indicadores de los servicios correspondientes

Demostrar la validez de un método basado en datos de epidemiología genética para evaluar las necesidades de servicios equitativos y costoeficaces de tratamiento y prevención de las hemoglobinopatías. Se obtuvieron datos demográficos y sobre la prevalencia de las distintas variantes de genes causantes de hemoglobinopatías a partir de bases de datos en línea, de referencias y de artículos publicados. Se creó una base de datos epidemiológica mundial sobre las hemoglobinopatías por países, incluidos cinco indicadores prácticos de servicios para expresar las necesidades de atención (indicador 1) y de prevención (indicadores 2 a 5).

Las hemoglobinopatías representan un importante problema sanitario en un 71% de los 229 países considerados, y en ese 71% se producen el 89% de todos los nacimientos. Cada año nacen más de 330 000 niños afectados (83% de casos de anemia de células falciformes y 17% de casos de talasemia).

Las hemoglobinopatías causan aproximadamente un 3,4% de las defunciones entre los niños menores de 5 años. A nivel mundial, en torno a un 7% de las mujeres embarazadas son portadoras de talasemia β o α cero, o de hemoglobina S, C, D Punjab o E, y más de un 1% de las parejas corren riesgo. Se debería informar a los portadores y a las parejas en riesgo de ese peligro y de las opciones para mitigarlo. El cribado de las hemoglobinopatías debería formar parte de los servicios básicos de salud en la mayoría de los países.

ملخص

الوبائيات العالمية لاضطرابات الهيموغلوبين ومؤشِّرات الخدمات المستمدة من واقعها

توضيح طريقة لاستخدام المعطيات الوبائية الجينية، لتقييم الاحتياجات من الخدمات العادلة والعالية المردود لمعالجة اضطرابات الهيموغلوبين والوقاية منها. حصل الباحثون على معطيات حول السمات الديمغرافية ومعدلات انتشار الضروب الجينية المسؤولة عن اضطرابات الهيموغلوبين وذلك من قواعد المعطيات المتاحة على الإنترنت في المصادر المرجعية ومن المقالات المنشورة، وأسسوا قاعدة معطيات عالمية لوبائيات اضطرابات الهيموغلوبين وفقاً لتوزيعها في البلدان، وهي تتضمَّن خمسة مؤشِّرات للخدمات العملية لتوضيح الاحتياجات للرعاية (المؤشِّر رقم 1) وللوقاية (المؤشِّرات من 2 إلى 5).

تمثِّل اضطرابات الهيموغلوبين مشكلة صحية جسيمة في 71 بلداً من بين مجمل البلدان التي يبلغ عددها 229 والتي يحتمل أن يصاب في هذه البلدان الـ 71، 89% من الولادات. ففي كل عام يولد 330 000 مولود مصاب باضطرابات الهيموغلوبين (83% منهم باضطرابات الخلية المنجلية، 17% بالثلاسيمية)؛ وتسبِّب اضطرابات الهيموغلوبين 3.4% من وفيات الأطفال الذين تقل أعمارهم عن 5 سنوات، وعلى الصعيد العالمي يحمل 7% تقريباً من الأمهات الحوامل بي أو ألف ثلاسيمية أو الهيموغلوبين S أو C أو D بنجاب أو E، كما يتعرَّض لخطر الإصابة أكثر من 1% من الأزواج. ينبغي توعية الحَمَلَة والأزواج المعرَّضين لاختطار مرتفع، عن ما هم معرضون له من خطر وعن الاختيارات المتاحة لهم لتقليل الخطر. وينبغي أن يكون تحرِّي اضطرابات الهيموغلوبين جزءاً من الخدمات الصحية الأساسية في معظم البلدان.

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