

# Modell's

Haemoglobinopathologist's

# Almanac

## Epidemiological Estimates for Haemoglobin Disorders: The World by WHO Region

### Comprising the following charts and tables:

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- Chart 2: Annual births
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Charts and tables compiled showing conservative estimates based on best available data presented on the World Wide Web in May 2007 by Professor Bernadette Modell (b.modell@pcps.ucl.ac.uk) and Matthew Darlison (m.darlison@ucl.ac.uk), UCL Centre for Health Informatics and Multiprofessional Education (CHIME) and World Health Organisation Collaborating Centre for the Community Control of Hereditary Disorders.

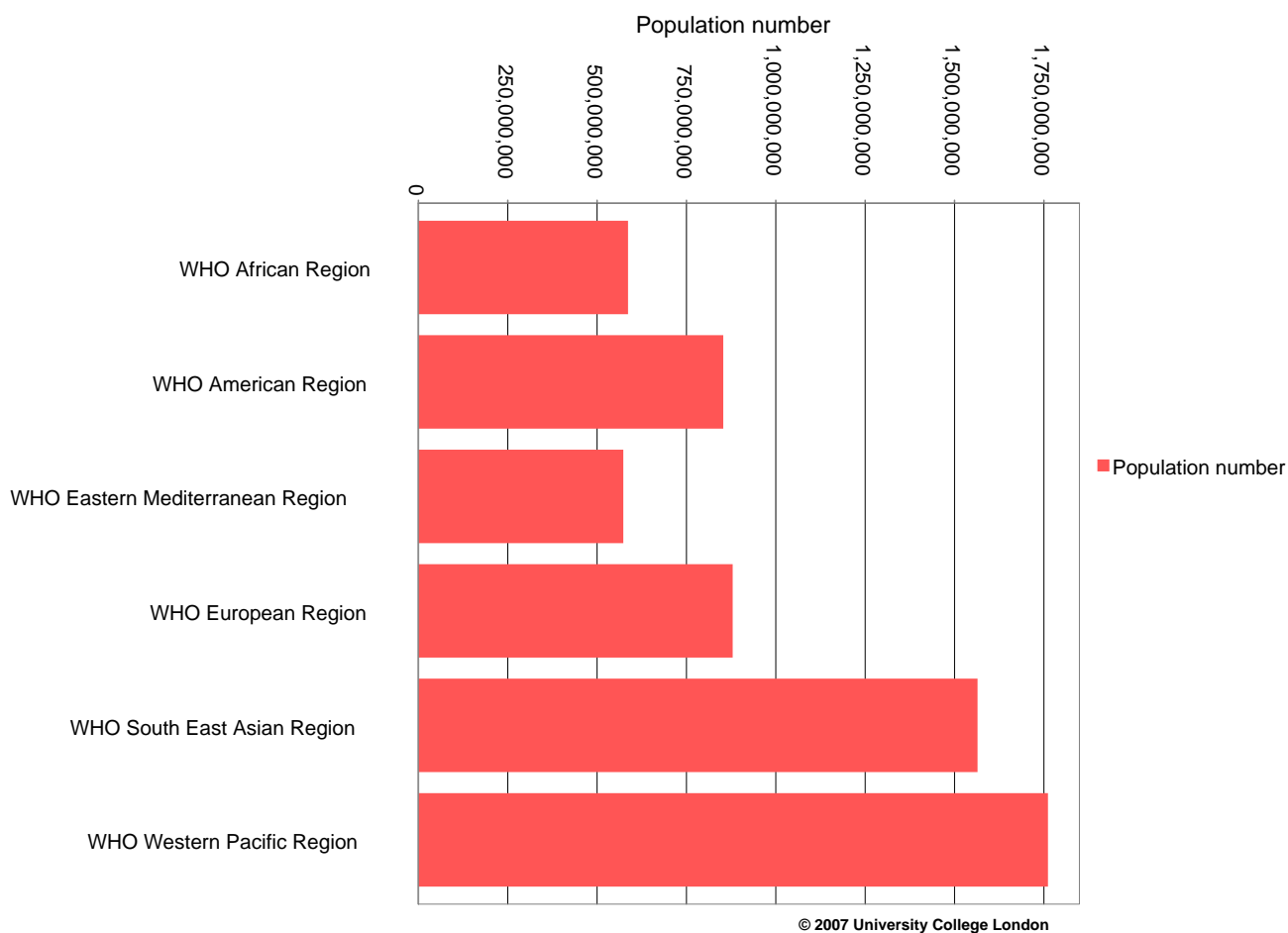
### Definitions used

In terms of alpha globin genotypes, we define as pathological homozygous alpha zero thalassaemia (causes hydrops fetalis) and the combination of alpha zero and alpha plus thalassaemia (causes haemoglobin H disease). Homozygous alpha plus thalassaemia is not considered to cause pathology. In terms of beta globin genotypes, we define as pathological haemoglobin S/C, homozygous haemoglobin S (SS), haemoglobin S/beta thalassaemia, homozygous beta thalassaemia, haemoglobin E/beta thalassaemia and some rare combinations. We define as harmless homozygous haemoglobin C (CC), haemoglobin C/beta thalassaemia, homozygous haemoglobin E (EE), homozygous haemoglobin D (DD), haemoglobin D/beta thalassaemia, and some other rare combinations.

WHO Regions are constituted as follows:

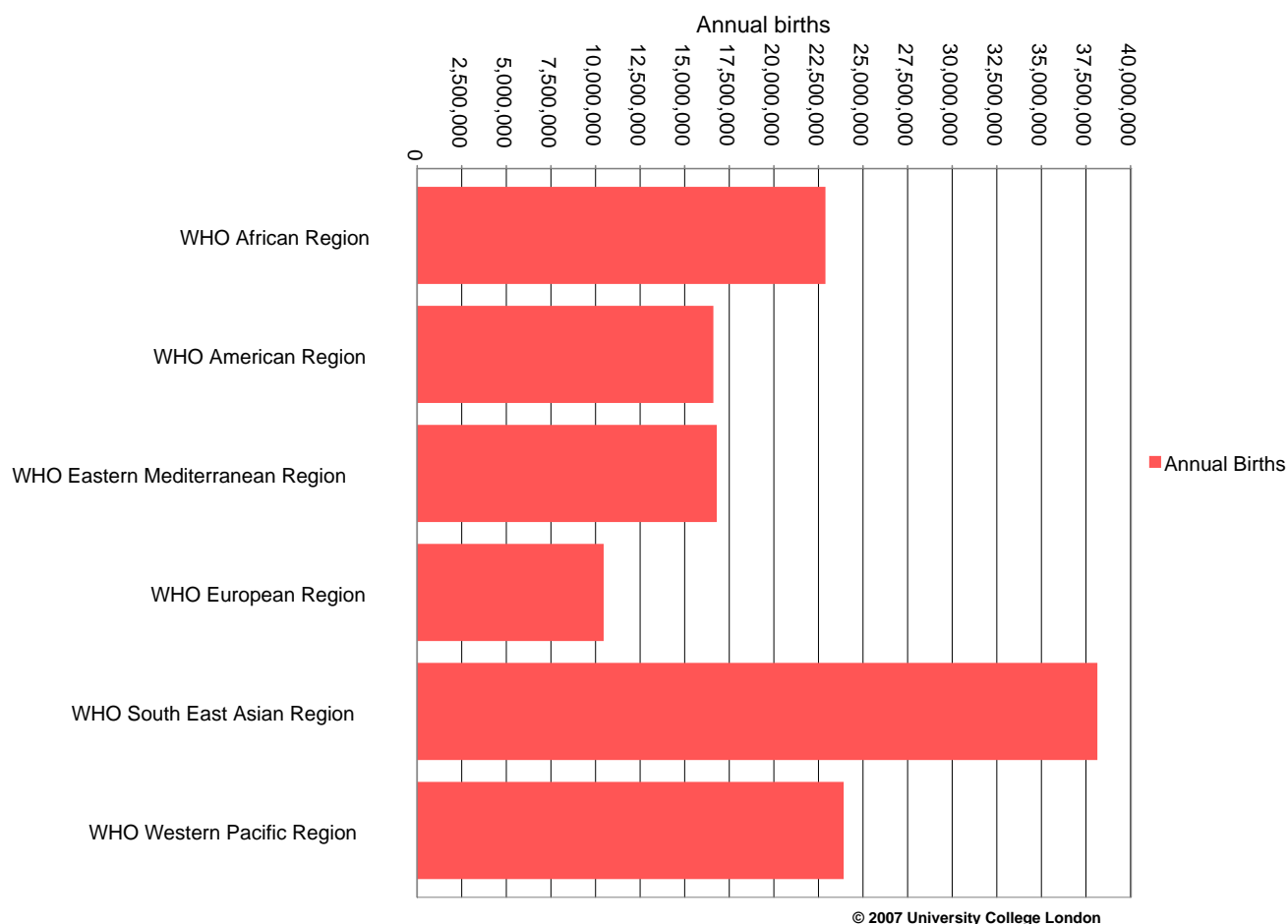
African Region (AFR)	American Region (AMR)	Eastern Mediterranean Region (EMR)	European Region (EUR)	South East Asian Region (SEAR)	Western Pacific Region (WPR)
<b>Northern Africa:</b> Algeria <b>Western Africa:</b> Benin, Burkina Faso, Cote d'Ivoire, Cape Verde, Gambia, Guinea-Bissau, Ghana, Guinea, Liberia, Mali, Mauretania, Niger, Nigeria, Sierra Leone, Saint Helena, Senegal, Togo <b>Middle Africa:</b> Angola, Central African Republic, Cameroon, Chad, Congo, Democratic Republic of the Congo (formerly Zaire), Equatorial Guinea, Gabon, Sao Tome and Principe <b>Eastern Africa:</b> Burundi, Comoros, Kenya, Madagascar, Malawi, Mauritius, Mayotte, Mozambique, Reunion, Rwanda, Seychelles, United Republic of Tanzania, Uganda, Zambia, Zimbabwe <b>Southern Africa:</b> Botswana, Lesotho, Namibia, South Africa, Swaziland	<b>Northern America:</b> Bermuda, Canada, Greenland, Saint Pierre and Miquelon, United States <b>Central America:</b> Belize, Costa Rica, El Salvador, Guatemala, Honduras, Mexico, Nicaragua, Panama <b>Caribbean:</b> Anguilla, Antigua and Barbuda, Aruba, Bahamas, Barbados, British Virgin Islands, Cayman Islands, Cuba, Dominica, Dominican Republic, Grenada, Guadeloupe, Haiti, Jamaica, Martinique, Montserrat, Netherlands Antilles, Puerto Rico, Saint Kitt's and Nevis, Saint Vincent and the Grenadines, Saint Lucia, Trinidad and Tobago, Turks and Caicos Islands, US Virgin Islands <b>South America:</b> Argentina, Bolivia, Brazil, Chile, Colombia, Ecuador, Falkland Islands, French Guiana, Guyana, Paraguay, Peru, Suriname, Uruguay, Venezuela	<b>Northern Africa:</b> Libyan Arab Jamahiriya, Morocco, Sudan, Tunisia, Western Sahara <b>Eastern Africa:</b> Djibouti, Egypt, Eritrea, Ethiopia, Somalia <b>Western Asia:</b> Bahrain, Cyprus, Iraq, Jordan, Kuwait, Lebanon, Oman, Qatar, Saudi Arabia, Syrian Arab Republic, United Arab Emirates, Yemen <b>South Central Asia:</b> Afghanistan, Iran, Islamic Republic of, Pakistan, Occupied Palestinian territory	<b>Northern Europe:</b> Channel Islands, Denmark, Estonia, Faroe Islands, Finland, Isle of Man, Iceland, Ireland, Latvia, Lithuania, Norway, Sweden, United Kingdom <b>Western Europe:</b> Austria, Belgium, France, Germany, Liechtenstein, Luxembourg, Monaco, Netherlands, Switzerland <b>Southern Europe:</b> Albania, Andorra, Bosnia and Herzegovina, Croatia, Gibraltar, Greece, Italy, The former Yugoslav Republic of Macedonia, Malta, Portugal, San Marino, Slovenia, Spain, Yugoslavia <b>Eastern Europe:</b> Belarus, Bulgaria, Czech Republic, Hungary, Republic of Moldova, Poland, Romania, Russian Federation, Slovakia, Ukraine <b>South Central Asia:</b> Kazakhstan, Kyrgyzstan, Tajikistan, Turkmenistan, Uzbekistan <b>Western Asia:</b> Armenia, Azerbaijan, Georgia, Israel, Turkey	<b>South Central Asia:</b> Bangladesh, Bhutan, India, Maldives, Nepal, Sri Lanka <b>South Eastern Asia:</b> East Timor, Indonesia, Myanmar, Thailand	<b>Eastern Asia:</b> China, China - Hong Kong, Japan, Korea, Democratic Peoples' Republic of Korea, Republic of, China - Macau, Mongolia, Taiwan, Province of China <b>South Eastern Asia:</b> Brunei Darussalam, Cambodia, Heard Island and McDonald Islands, Lao Peoples' Democratic Republic, Malaysia, Philippines, Singapore, Vietnam <b>Melanesia, Micronesia, Polynesia:</b> American Samoa, Cook Islands, Fiji, French Polynesia, Guam, Kiribati, Marshall Islands, Micronesia, Federated States of New Caledonia, Northern Mariana Islands, Nauru, Niue, Palau, Pitcairn, Papua New Guinea, Samoa, Solomon Islands, Tokelau, Tonga, Tuvalu, Vanuatu, Wallis and Futuna Islands <b>Australia and New Zealand:</b> Australia, New Zealand

Chart 1: Population



<i>WHO Region</i>	<i>Population number</i>
WHO African Region	586,362,800
WHO American Region	852,768,758
WHO Eastern Mediterranean Region	573,248,538
WHO European Region	879,191,011
WHO South East Asian Region	1,564,232,000
WHO Western Pacific Region	1,761,269,066
The World population total	6,217,072,173

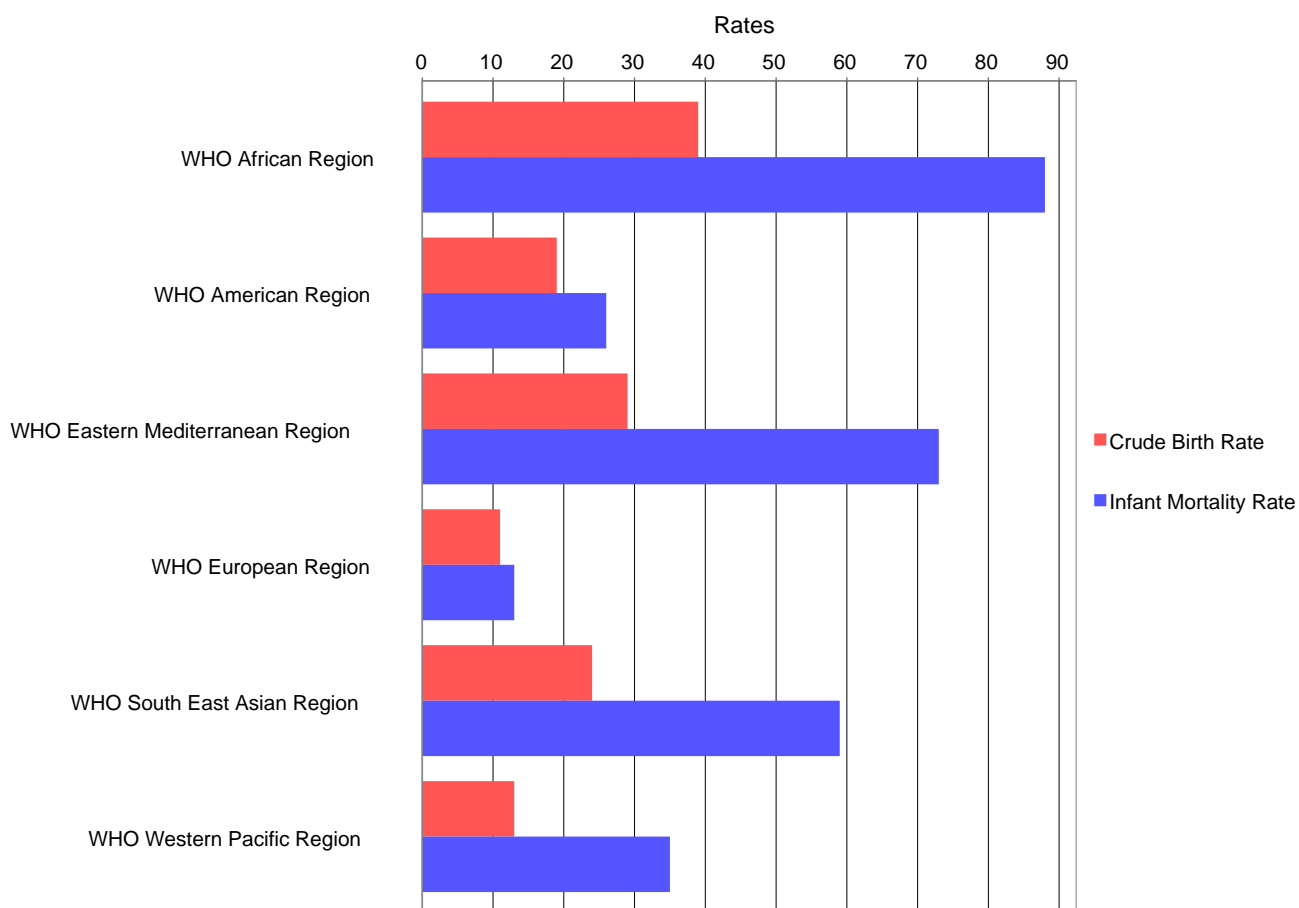
Chart 2: Annual births



Indicates requirement for carrier screening during pregnancy. The annual number of people marrying (not shown) is the indicator for pre-marital or pre-pregnancy screening.

WHO Region	Annual Births
WHO African Region	22,894,989
WHO American Region	16,608,920
WHO Eastern Mediterranean Region	16,798,470
WHO European Region	10,456,353
WHO South East Asian Region	38,138,978
WHO Western Pacific Region	23,914,239
The World total births	128,811,949

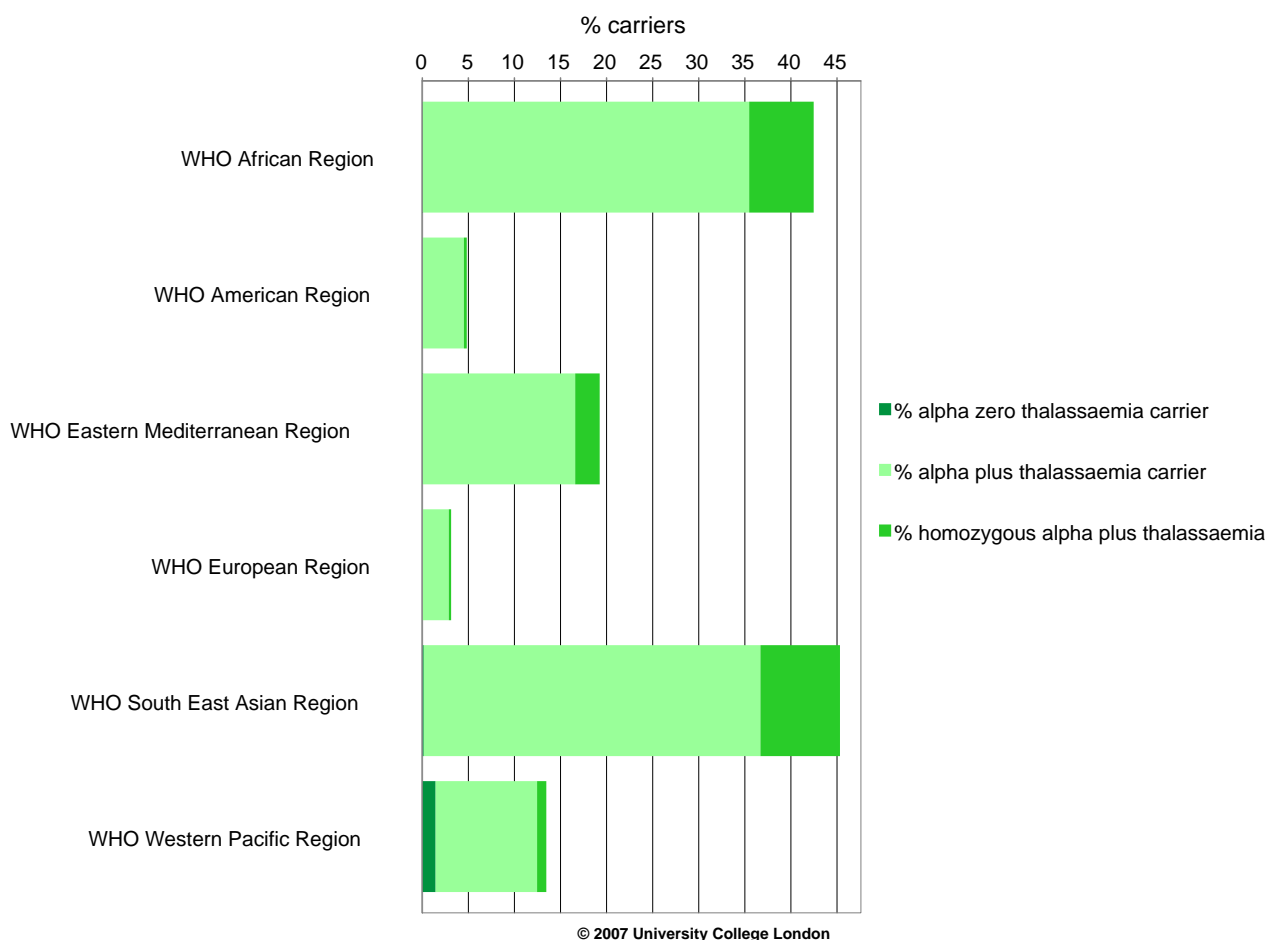
Chart 3: Crude birth rate and infant mortality rate



Crude birth rate (CBR) = live births per 1000 population per year; infant mortality rate (IMR) = deaths under 1 year, per 1,000 live births.

WHO Region	Crude Birth Rate	Infant Mortality Rate
WHO African Region	39	88
WHO American Region	19	26
WHO Eastern Mediterranean Region	29	73
WHO European Region	11	13
WHO South East Asian Region	24	59
WHO Western Pacific Region	13	35
The World aggregate rates	20	53

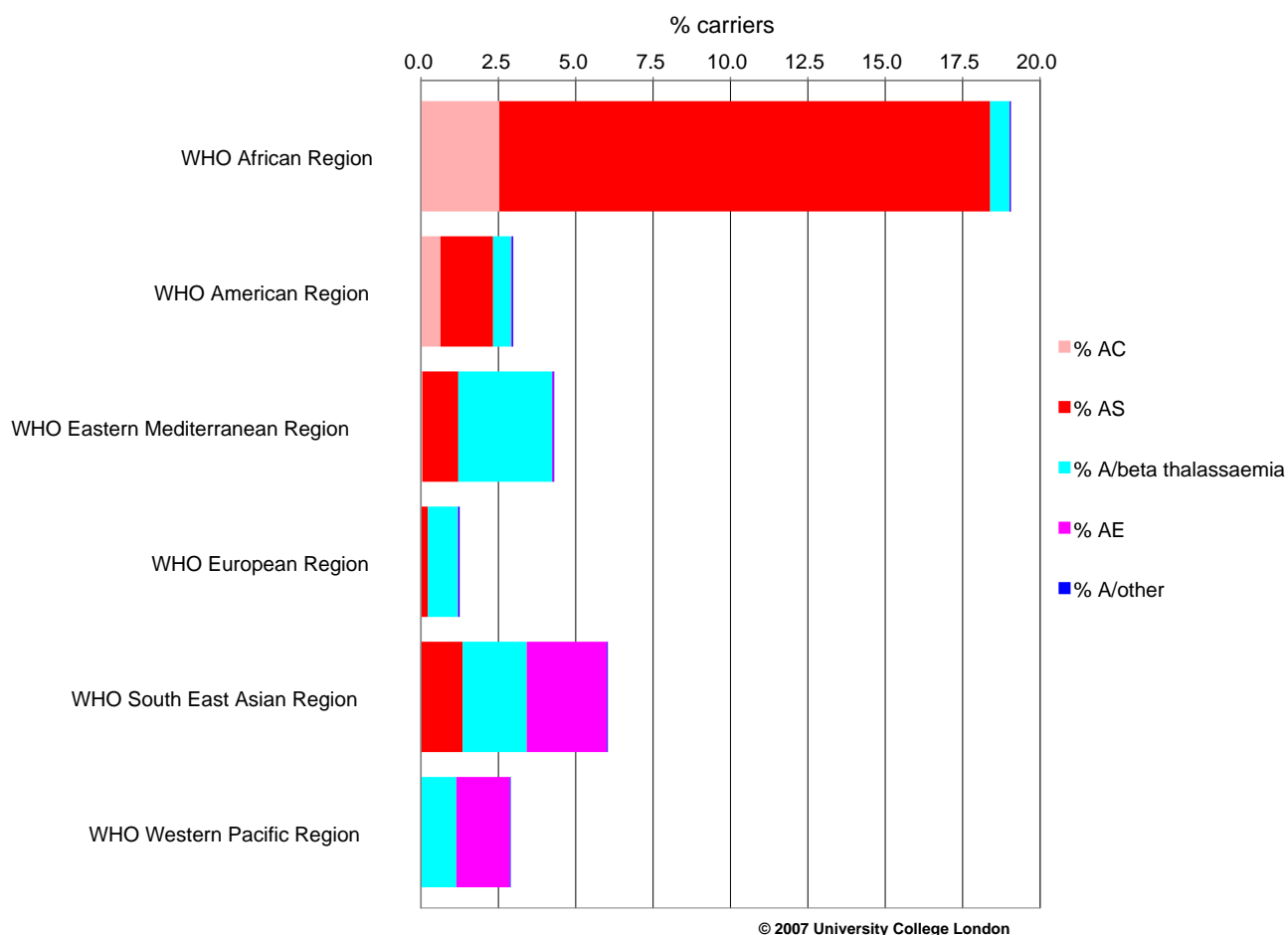
Chart 4: Per cent of pregnant women carrying alpha globin gene variants, by variant



Data is for deletional alpha globin gene variants. Homozygous alpha plus thalassaemia is included as it is common and harmless.

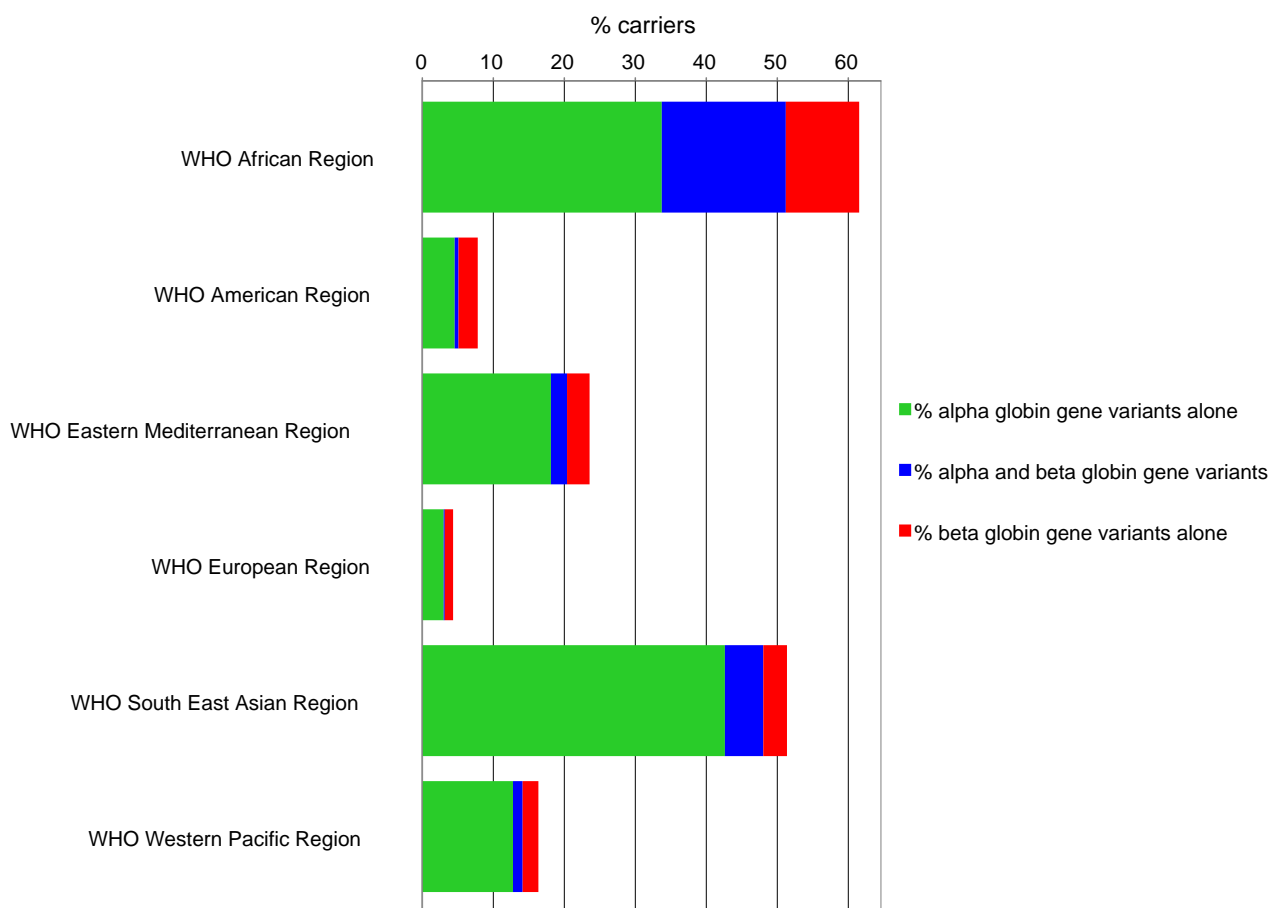
WHO Region	% alpha zero thalassaemia carrier	% alpha plus thalassaemia carrier	% homozygous alpha plus thalassaemia
WHO African Region	0	35.468	7.001
WHO American Region	0.045	4.47	0.315
WHO Eastern Mediterranean Region	0.001	16.595	2.658
WHO European Region	0.009	2.868	0.25
WHO South East Asian Region	0.157	36.531	8.636
WHO Western Pacific Region	1.444	11.012	0.999
The World aggregate carrier rates	0.321	22.138	4.394

Chart 5: Per cent of pregnant women carrying beta globin gene variants, by variant



WHO Region	% AC	% AS	% A/beta thalassaemia	% AE	% A/other
WHO African Region	2.531	15.861	0.64	0.001	0.03
WHO American Region	0.631	1.699	0.585	0.036	0.03
WHO Eastern Mediterranean Region	0.045	1.16	3.028	0.042	0.03
WHO European Region	0.02	0.202	0.967	0.021	0.04
WHO South East Asian Region	0	1.345	2.07	2.595	0.03
WHO Western Pacific Region	0	0.001	1.141	1.726	0.03
The World aggregate carrier rates	0.539	3.604	1.487	1.101	0.031

Chart 6: Per cent of pregnant women carrying any haemoglobin gene variant

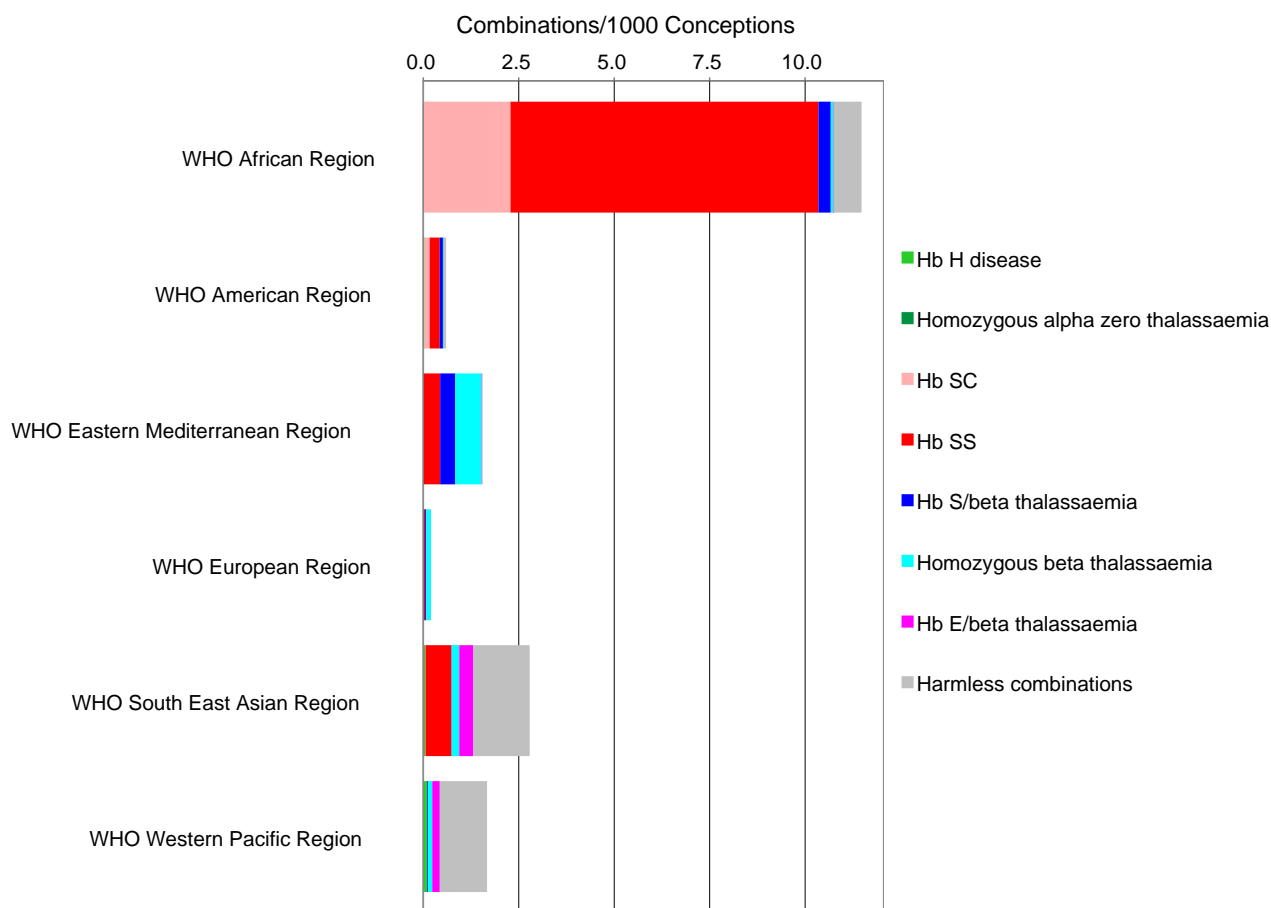


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Alpha and beta globin gene variants are inherited independently and do not interact to cause disease: many people carry both alpha and beta variants.

WHO Region	% carriers of alpha globin gene variants alone	% carriers of both alpha and beta globin gene variants	% carriers of beta globin gene variants alone	Total % carriers
WHO African Region	33.728	17.482	10.321	61.531
WHO American Region	4.56	0.541	2.711	7.812
WHO Eastern Mediterranean Region	18.121	2.265	3.173	23.559
WHO European Region	3.012	0.156	1.171	4.339
WHO South East Asian Region	42.621	5.407	3.335	51.363
WHO Western Pacific Region	12.776	1.36	2.218	16.354
The World aggregate carrier rates	24.182	5.338	4.092	33.612

Chart 7: Conceptions/1000 with combinations of significant variants



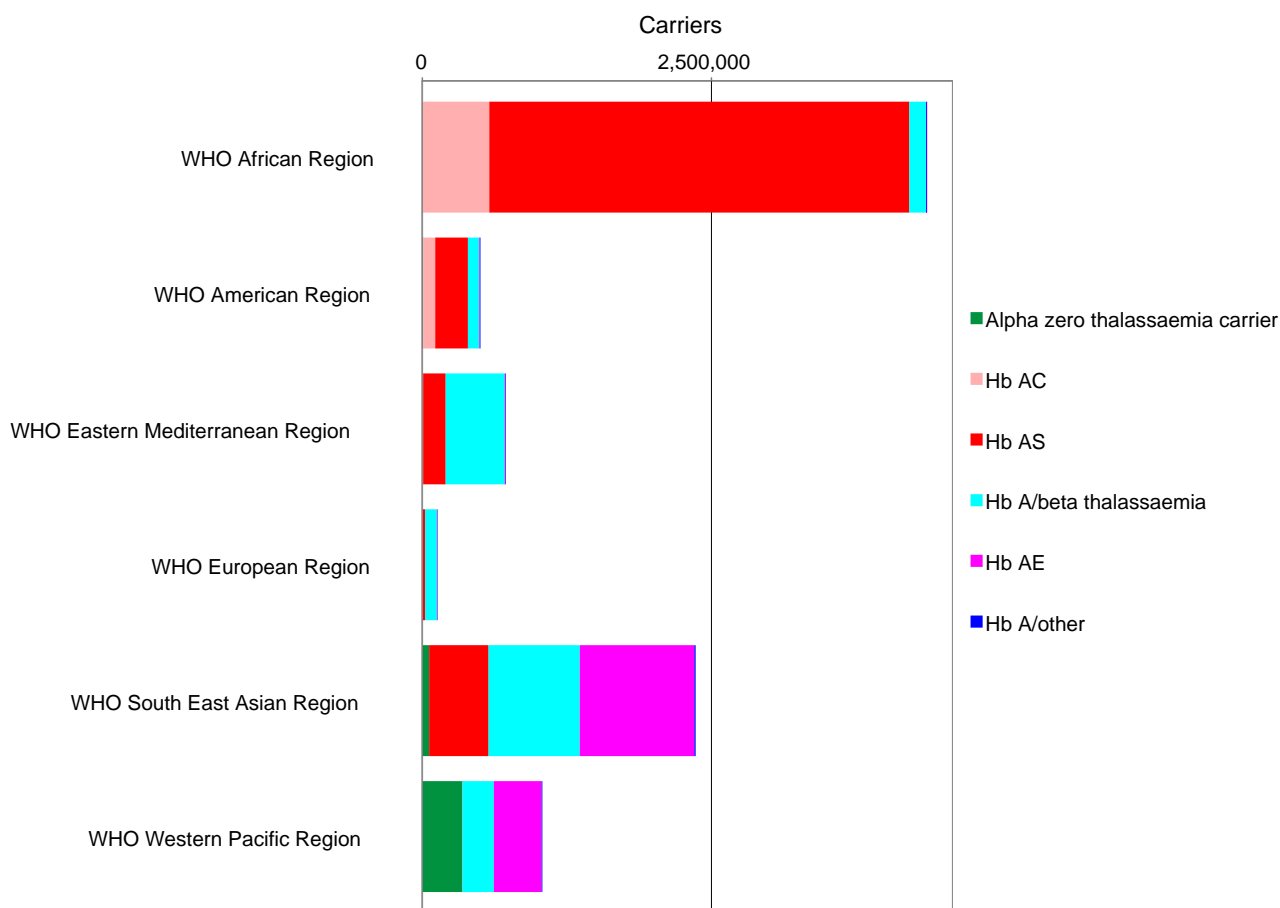
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Pathological combinations are shown individually. Harmless combinations are grouped together. For details see definitions.

WHO Region	Hb H disease	Homozygous alpha zero thalassaemia	Hb SC	Hb SS	Hb S/beta thalassaemia	Homozygous beta thalassaemia	Hb E/beta thalassaemia	Harmless combinations	Total affected conceptions	Total conceptions
WHO African Region	0	0	2.283	8.066	0.322	0.066	0	0.74	10.737	11.477
WHO American Region	0.029	0.001	0.136	0.265	0.083	0.021	0.005	0.058	0.54	0.597
WHO Eastern Mediterranean Region	0	0	0.004	0.439	0.395	0.688	0.009	0.019	1.536	1.554
WHO European Region	0	0	0.012	0.036	0.026	0.125	0.001	0.006	0.2	0.206
WHO South East Asian Region	0.059	0.002	0	0.676	0.007	0.198	0.371	1.474	1.312	2.786
WHO Western Pacific Region	0.108	0.007	0	0	0	0.12	0.198	1.238	0.433	1.671
The World aggregate rates	0.041	0.002	0.425	1.728	0.124	0.195	0.148	0.808	2.663	3.471



Chart 8: Annual pregnant carriers of significant variants

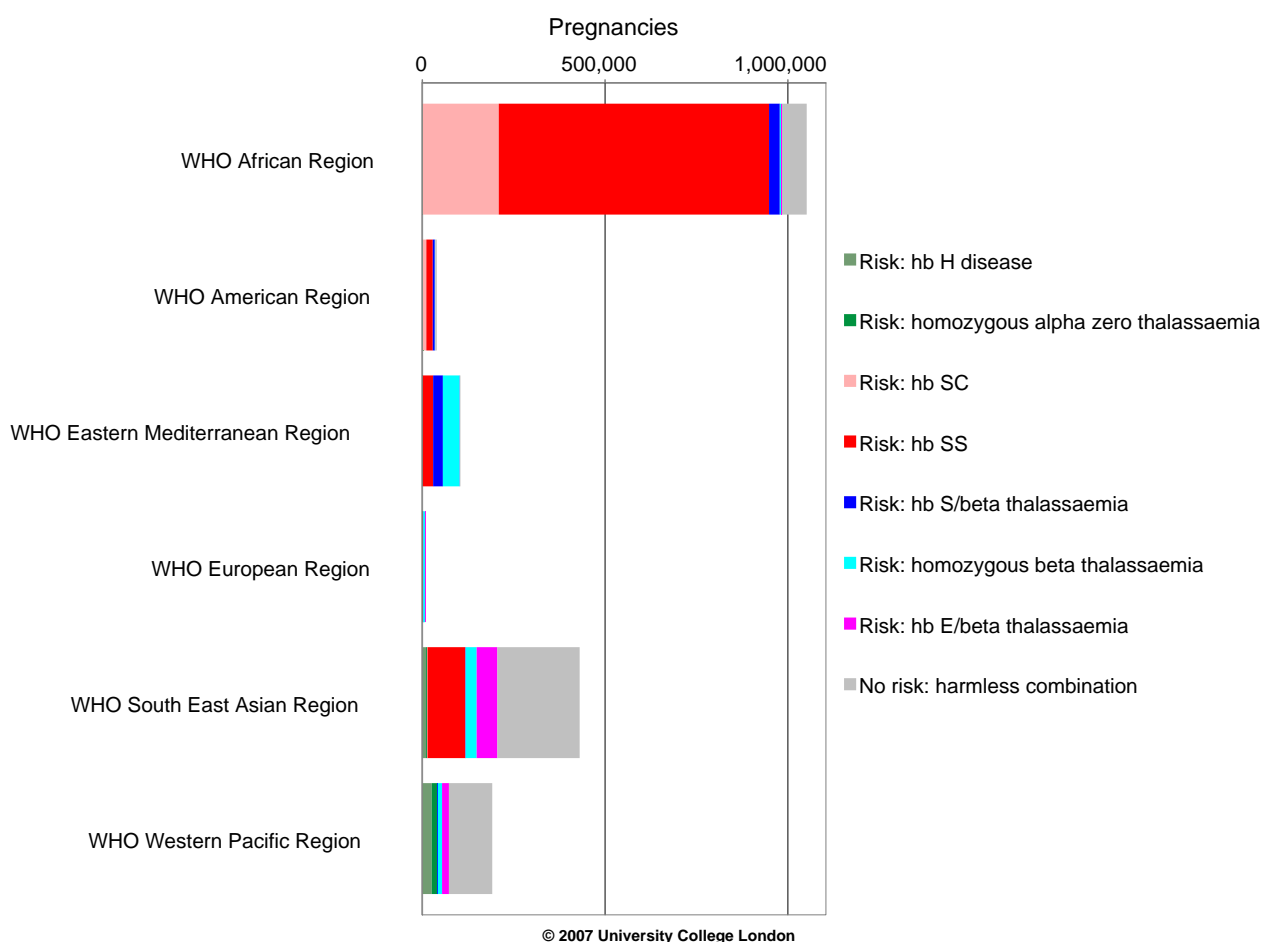


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Indicates requirement for information for carriers, and the offer of partner testing. Also indicates annual carriers detectable by neonatal screening.

WHO Region	Alpha zero thalassaemia carrier	Hb AC	Hb AS	Beta thalassaemia carrier	Hb AE	Hb A/other	Total carriers
WHO African Region	86	579,472	3,631,380	146,420	117	6,842	4,364,317
WHO American Region	7,489	104,717	282,193	97,194	6,057	4,963	502,613
WHO Eastern Mediterranean Region	89	7,542	194,895	508,667	7,043	5,025	723,261
WHO European Region	966	2,078	21,071	101,046	2,155	4,154	131,470
WHO South East Asian Region	59,835	0	512,800	789,328	989,599	11,452	2,363,014
WHO Western Pacific Region	345,427	23	328	272,906	412,655	7,098	1,038,437
The World total pregnant carriers	413,892	693,832	4,642,667	1,915,561	1,417,626	39,534	9,123,112

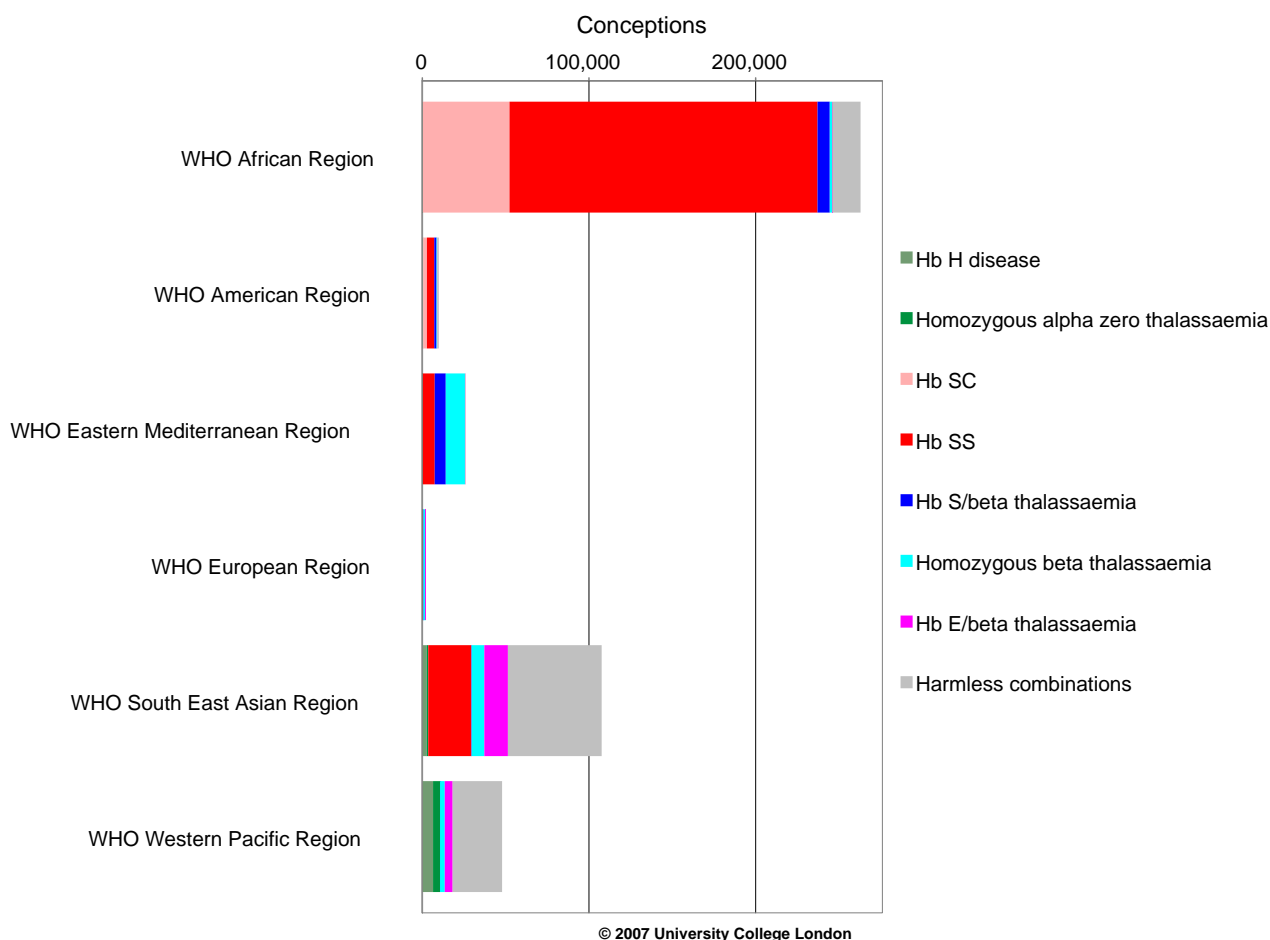
Chart 9: Annual pregnancies where both parents carry a significant variant



Indicates requirement for expert risk assessment, genetic counselling and the offer of prenatal diagnosis.

WHO Region	Risk: hb H disease	Risk: homozygous alpha zero thalassaemia	Risk: hb SC	Risk: hb SS	Risk: hb S/beta thalassaemia	Risk: homozygous beta thalassaemia	Risk: hb E/beta thalassaemia	No risk: harmless combination	Total at-risk pregnancies	Total pregnancies requiring risk assessment
WHO African Region	40	0	209,152	739,168	29,384	6,004	8	67,740	983,756	1,051,496
WHO American Region	1,932	64	9,064	17,656	5,456	1,340	304	3,820	35,816	39,636
WHO Eastern Mediterranean Region	4	0	240	29,520	26,508	46,216	632	1,244	103,120	104,364
WHO European Region	0	0	484	1,468	1,068	5,172	36	228	8,228	8,456
WHO South East Asian Region	10,396	4,068	0	103,068	1,076	30,208	56,604	225,000	205,420	430,420
WHO Western Pacific Region	25,900	16,600	4	32	8	11,440	18,928	118,488	72,912	191,400
The World total pregnancies	38,272	20,732	218,944	890,912	63,500	100,380	76,512	416,520	1,409,252	1,825,772

Chart 10: Annual conceptions with combinations of significant gene variants



Indicates (potential) annual affected births, and annual increase in requirements for patient care. Baseline for assessing effects of prevention.

WHO Region	Hb H disease	Homozygous alpha zero thalassaemia	Hb SC	Hb SS	Hb S/beta thalassaemia	Homozygous beta thalassaemia	Hb E/beta thalassaemia	Harmless combinations	Total affected conceptions	Total conceptions with combinations
WHO African Region	10	0	52,288	184,792	7,346	1,501	2	16,935	245,939	262,874
WHO American Region	483	16	2,266	4,414	1,364	335	76	955	8,954	9,909
WHO Eastern Mediterranean Region	1	0	60	7,380	6,627	11,554	158	311	25,780	26,091
WHO European Region	0	0	121	367	267	1,293	9	57	2,057	2,114
WHO South East Asian Region	2,599	1,017	0	25,767	269	7,552	14,151	56,250	51,355	107,605
WHO Western Pacific Region	6,475	4,150	1	8	2	2,860	4,732	29,622	18,228	47,850
The World total pregnancies	9,568	5,183	54,736	222,728	15,875	25,095	19,128	104,130	352,313	456,443