Haemoglobinopathies: current situation of genetic counselling and diagnosis
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Haemoglobinopathies are inherited disorders of haemoglobin. With 7% of the global population being carriers and 300,000–400,000 affected children born each year worldwide, these are the most common monogenic disorders in humans. The term “haemoglobinopathies” refers to two main groups of disorders: 1) thalassaemias and 2) haemoglobin variants. Haemoglobinopathies are usually transmitted in an autosomal recessive manner, which means that heterozygous individuals are almost always asymptomatic carriers, whereas homozygous or compound heterozygous individuals have symptoms of varying degrees of severity. Haemoglobinopathies have been recognized as a public health priority by the World Health Organization, but in many European countries, these diseases remain under recognized. Relevant preventive measures, education, information, diagnosis, and clinical care are not part of a comprehensive health programme.
Primary prevention i.e. testing for haemoglobinopathies before conception, or at least antenatal screening, is recommended for all individuals at risk. In continental Europe, restrictive or no policies are encountered most often. Primary prevention is related to genetic counselling. Patients or relatives, at risk of an inherited disorder, are advised of the consequences and nature of the disorder, the probability of developing or transmitting it, the options open to them in management and family planning in order to prevent, avoid or ameliorate it. Genetic counselling is a complex process including supportive and diagnostic aspects. Genetics counsellors are acutely aware of the anxiety engendered by positive screening or diagnostic tests in pregnancy, they are also aware of the difficulties of giving information to families with a different cultural, emotional and religious background. Secondary prevention has a main objective: to detect newborn infants with a sickle cell disorder and to provide them with early treatment and care. In United Kingdom screening of newborns has been linked with an antenatal screening programme.
For example, the evaluation of the efficiency of prenatal diagnosis and neonatal screening for haemoglobinopathies in Brussels will be presented. The conclusion of this evaluation was that prenatal screening is currently not efficient enough. It is too often performed too late in the pregnancy or not at all. Even if prenatal diagnosis is declined, women, couple with a haemoglobinopathy value the opportunity to receive information about their risk status.
In the field of genetic counselling for haemoglobinopathies, many challenges remain to be addressed and not only in Belgium.