Sickle cell disease

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Sickle cell disease (SCD) is a group of inherited blood disorders that cause severe pain, reduce life expectancy and require significant self-management, but are often associated with stigma and discrimination. They involve abnormal forms of haemoglobin that polymerize, or gel, when deoxygenated, making the red blood cells stiffen and elongate into a sickle or crescent shape. This disrupts the circulation, starving the affected tissue and organs of oxygen and causing very severe acute pain. The spleen, a vital part of the immune system, is very vulnerable to sickling, so people with SCD are susceptible to infections as well as chronic anaemia and a range of life-threatening complications (Ashley-Koch et al., 2000).

SCD is caused by abnormal forms of the beta polypeptide chain gene on chromosome 11. Inheritance is autosomal and recessive, so abnormal genes must be inherited from both parents. People who inherit an abnormal gene from just one parent are trait carriers and usually have almost no symptoms. The sickle trait provides resistance against malaria (Bunn, 2013), which is why SCD mainly affects those who live or have family origins in regions where malaria has been endemic (Williams & Obaro, 2011).

Over 300,000 babies are born with SCD every year worldwide; prevalence is highest in sub-Saharan Africa, but is also high in India and the Middle East, and in high prevalence areas the trait carrier rate ranges from 5% to 40% (Piel et al., 2013). There are about 100,000 people with SCD in the USA (Hassell, 2010) and over 12,000 in the UK (Streetly et al., 1997). Based on new-born screening, the incidence of SCD in England was 0.54 per 1,000, but ranged from none per 1,000 in Dorset and Somerset to 3.05 per 1,000 in South East London. Trait carrier rates were 15 per 1,000 for England, but ranged from 1.85 per 1,000 for babies recorded as 'White British', to 145 per 1,000 for babies recorded as 'Black African' (Streetly et al., 2010).

Survival rates have improved significantly over the last 50 years, and in 2005 the median age of death for people with SCD in the US was 38 years for males and 42 years for females (Lanzkron et al., 2013). Fewer people with SCD survive to adulthood in developing countries, however, where better screening and counselling programmes are needed (Aneke & Okocha, 2016), and where SCD is often associated with significant stigma (Ola et al., 2016).

Preconception screening aims to identify SCD trait carriers so they can be offered genetic counselling and make informed reproductive choices, and is recommended for everyone whose family origins indicate increased risk. Antenatal screening is recommended for all pregnancies in high prevalence areas, and for pregnancies in low prevalence areas where either parent's family origins indicate increased risk. New-born screening aims to identify babies with SCD so that comprehensive care and protection against infection and other complications can begin as early as possible, and is recommended for all new-born babies (Ryan et al., 2010).

Bone marrow transplantation can potentially cure SCD, but relatively few operations take place because transplantation is expensive and risky and there are limited donors (Walters, 2015). Current evidence-based management for SCD includes preventative antibiotics for children, blood transfusions, hydroxyurea, and tests and treatments for a range of complications (National Institutes of Health, National Heart, Lung, and Blood Institute, 2014). Hydroxyurea can reduce painful episodes and other complications but its benefits are not yet fully certain (Brawley et al., 2008). Producing evidence-based treatment recommendations has raised questions about how well they are followed by healthcare providers, and about patients' access to quality healthcare (Adams-Graves & Bronte-Jordan, 2016).

Managing acute painful sickling episodes is a key element of treatment, and guidelines in both the UK and US emphasise the importance of prompt and effective analgesia (National Institute for Health and Care Excellence, 2015; National Institutes of Health, National Heart, Lung, and Blood Institute, 2014). Aggressive treatment of sickling pain is justified by the phases and mechanisms of painful sickling episodes, for undertreated acute sickling pain can lead to more serious tissue damage and chronic pain syndromes (Ballas et al., 2012). However, there is plenty of evidence of undertreated SCD pain, often associated with negative attitudes and beliefs on the part of healthcare providers about the condition and the people affected (Haywood et al., 2013; Labbé et al., 2005; GLassberg et al., 2013). The misperception of pain behaviours as symptoms of addiction is one way in which negative attitudes and beliefs can lead to undertreated pain (Elander et al., 2004; 2006). However, interventions can improve healthcare providers' attitudes and beliefs (Haywood et al., 2011), including in emergency departments (Freiermuth et al., 2016).

People can manage SCD better if they are in skilled occupations, and more education was associated with less need for unplanned healthcare related to SCD (Jonassaint et al., 2016), so education is extremely important. Children with SCD report significant difficulties at school, however (Dyson et al., 2010a), and stigmatization begins early as many affected children and adolescents decide not to disclose their condition (Dyson et al., 2010b). Education and cognitive ability may also be affected by neuropsychological impairments caused by sickling episodes in the brain, including 'silent' or unnoticed cerebral infarcts (Wang et al., 2001).

Adolescent transitions are difficult for young people with SCD, because smoking, drinking alcohol, exertion and exposure to cold and wet are risk factors for sickling episodes. The adolescent and young adult years are associated with increased needs for medical treatment, and the transition from paediatric to adult hospital care can be difficult for young people with SCD and should be actively managed by health care providers (Treadwell et al., 2011). Quality of life is lower for people with SCD compared with the general population (McClish et al., 2005), and psychological interventions aiming to improve adjustment and quality of life include those based on cognitive-behavioural therapy (Anie et al., 2002) and acceptance and commitment therapy (Masuda et al., 2011).

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