Haemophilia

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Haemophilia A and haemophilia B are inherited bleeding disorders caused by deficiencies in blood clotting factor proteins. They are sex-linked recessive disorders that are usually transmitted from carrier mother to affected son. Factor VIII deficiency causes haemophilia A, which affects about one in 5,000 males, and factor IX deficiency causes haemophilia B, which affects about one in 30,000 males (Kliegman, 2011). In 2012 there were estimated to be 6,742 people with haemophilia in the UK, 6,035 in France, 4,660 in Germany, and 18,628 in the USA (World Federation of Hemophilia, 2013). For people with haemophilia, spontaneous bleeding in joints, muscles and other soft tissues, as well as bleeding caused by injury or surgery, is not controlled by blood clotting, and repeated joint bleeds can damage the joints, leading to arthropathy, disability and chronic pain (Acharya, 2012).

Trials of gene therapy treatment show encouraging preliminary results but also significant limitations (Nienhuis et al., 2016), and replacement clotting factor is currently the main treatment. Prophylaxis (preventative clotting factor treatment) can reduce bleeds and improve quality of life (Royal et al., 2002) but prophylaxis is expensive (Zhou et al., 2015) and 20%-30% of people with haemophilia A develop inhibitors, which make replacement clotting factors less effective (Tabriznia-Tabrizi et al., 2016). Physical activity is also important, and exercise can improve joints and mobility (Mulvany et al., 2010; Schäfer et al., 2016).

Life expectancy for people with haemophilia has improved but is still reduced (Mazepa et al., 2016). During the 1970s and 1980s, many people with haemophilia were infected with HIV and hepatitis C through contaminated clotting factor concentrates, and the organisation *Tainted Blood* was set up "to support and to achieve justice for those infected and affected by contaminated blood and blood products in the United Kingdom" (https://www.taintedblood.info/tb/). One study showed that boys with hemophilia and HIV

infection showed considerable resilience in terms of adaptive behavior and emotional and behavioral problems (Nichols et al., 2000).

Inheritance raises difficult family issues, and genetic carrier testing for haemophilia is generally not recommended for children, but parents in one study were happy to know the results even when they showed that a child was a carrier (Vears et al., 2016). An ethnographic study of families affected by haemophilia showed that the main effects were on family relationships, school, employment and travel (Khair et al., 2014). Prophylaxis facilitated more regular family routines in one study, but did not completely normalize family life (Emiliani et al., 2011).

In other studies, behavioural techniques such as counterconditioning, distraction and differential positive reinforcement helped to improve a child's adherence to clotting factor treatment (Penica & Williams, 2008), and motivation and autonomy were key factors in increasing self-care for children with haemophilia (Bérubé et al., 2016). Attention to children's pain is important because qualitative research showed how pain affects children's experiences of haemophilia and treatment (Spitzer, 1993).

Physical quality of life is lower among adults with haemophilia than the general population, and lower among people with haemophilia who have more joint damage or are not treated with prophylaxis, but mental quality of life is less affected by haemophilia and less closely associated with joint status (Poon et al., 2012; Royal et al., 2002). Generic measures of quality of life, such as the SF-36, may not be specific or responsive enough to changes in health status (Szende et al., 2003), and there are haemophilia-specific measures of quality of life and subjective well-being (Remor et al., 2004; Remor, 2013).

The impact of pain for people with haemophilia, especially in developing countries, is beginning to be more widely recognised (Humphries & Kessler, 2016), and in a study covering over 5,000 adults with haemophilia at treatment centres in Europe, 67% had arthropathy and 35% had chronic pain (Holstein et al., 2012). A survey in the USA showed that 39% of people with haemophilia believed their pain was not well treated and many reported using alcohol and illicit drugs to manage pain (Witkop et al., 2012). Acute bleeding pain should be treated promptly with replacement clotting factor but chronic joint pain should not, so it is important to distinguish chronic joint pain from acute bleeding pain, which some people with haemophilia may have difficulty doing (Witkop et al., 2011).

In studies of the impact of pain on quality of life among people with haemophilia, pain intensity influenced physical quality of life and pain acceptance influenced mental quality of life, whereas active pain coping did not influence either physical or mental quality of life (Elander et al., 2009; Elander et al., 2013). In a DVD intervention to improve readiness to self-manage chronic joint pain caused by haemophilia, all the content was presented by five men with haemophilia who described their own experiences of living with joint pain, including its impact on their lives and how they had adjusted their life goals and values

accordingly (Elander et al., 2011). Compared with a written information booklet, the DVD increased readiness to self-manage pain, and improved mental health-related quality of life among those with only high school education (Elander et al., 2011; Stalker & Elander, 2015).

Evidence like this suggests that behavioural and psychological factors influence self-management and quality of life in the same ways among people with haemophilia as among those with other chronic pain conditions, so there should be scope for interventions developed for other chronic pain conditions to be adapted for people with haemophilia, provided that careful account is taken of the need to respond promptly to acute bleeding pain by administering clotting factor (Elander, 2014).

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