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Review

Down syndrome: An integrative review

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ABSTRACT

Down syndrome is a complex genetic disorder resulting in three copies of chromosome 21. Babies with this genetic disorder will have recognisable characteristic facial features that will differ from one baby to another. They will also have some degree of cognitive impairment and learning difficulties. There are many medical conditions associated with Down syndrome, however, due to recent medical advances there have been improvements in their health and longevity. This has led to a rise in people with Down syndrome developing Alzheimer's disease as they age.

The purpose of this review is to provide insight into the impacts that Down syndrome has on foetal development as well as ongoing health issues up to adulthood. There were many ethical issues raised surrounding the baby Doe case and will also be explored in this review. CINAHL (EBSCO) was the primary medical database for this review retrieving 147 results in relation to Down syndrome and foetal development. An additional search was made retrieving 12 results in relation to ethical issues surrounding prenatal diagnosis of Down syndrome. Further resources were used such as websites and neonatal nursing textbooks.

This paper aims to provide a snapshot of Down syndrome with consideration given to the short and long-term outcome for the baby, and the consequences for the growing child and his/her family. It is essential for neonatal nurses to understand the complexities of this genetic disorder, how to care for babies with Down syndrome, and how to provide support to parents and families.

Contents

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1. Introduction

Down syndrome is one of the most common genetic disorders that impacts foetal development, affecting 1 in every 1150 live births in Australia (Tracy, 2011). It is also known as Down's syndrome or trisomy 21 and was discovered by John Langdon Down in 1866. It is a chromosomal disorder where the individual has an additional copy of chromosome 21, either full or partial (Your Genome, 2015). Genetics plays a major role in foetal development and is where chromosomal abnormalities occur. Chromosomal abnormalities with Down syndrome occur during the meiotic cellular division phase (Perkins, 2017). This can happen due to increased maternal age, although there are mothers younger than 35 that have conceived a baby with Down syndrome (Perkins, 2017). It is important to have prenatal screening to monitor foetal development and to make an accurate diagnosis during this period Differences in culture and ethnicity can impact prenatal screening discussion and decision making, raising many ethical issues.

Over recent decades, the health and life expectancy of Australians with Down syndrome has improved resulting in more people living into adulthood (Tracy, 2011). Furthermore, there are a number of medical conditions associated with Down syndrome, the most common being cardiac defects, leukaemia, gastrointestinal issues, vision and hearing issues, dental issues, thyroid disease, obstructive sleep apnoea, epilepsy, and Alzheimer disease (Perkins, 2017). Surgical interventions may be required to correct any medical issues to improve baby's outcomes. Down syndrome is also one of the leading causes of intellectual disability (Asim et al., 2015). Neonatal nurses act as advocates for babies and their families. They need to be able to provide the emotional support as well as educate parents and families about Down syndrome (Kenner and Lott, 2014). It is also paramount for neonatal nurses to understand how Down syndrome impacts families as well as how to care for babies with this genetic disorder.

2. Methodology

Relevant research relating to Down syndrome and foetal development was identified by searching the health databases for primary research material. A total of five research databases were searched for this review; CINAHL (EBSCO), MEDLINE (OVID), PubMed, ProQuest Health & Medicine, and SCOPUS (Elsevier), although, key articles were obtained primarily from CINAHL (EBSCO). In order to ensure that relevant studies were not missed, the search terms remained broad. The terms were 'Down syndrome', 'pregnancy', and 'fetus/foetus' using a Boolean search type approximately 447 results were retrieved. From these results the search criteria were further filtered to approximate the results from the last ten years and were selected based on their peer-reviewed status. From this, 147 results were retrieved. The articles were then selected based upon their relevance to Down syndrome and foetal development and, where possible, Australian data was a preferred resource.

A further search was undertaken to explore the ethical issues surrounding prenatal diagnosis of Down syndrome. The terms were 'Down syndrome' and 'ethics', using a Boolean search type with 12 results retrieved. All 12 articles were reviewed and related to prenatal diagnosis. Articles selected dated back to the 1980s, although included due to their relevance to ethical issues surrounding prenatal diagnoses of Down syndrome.

A comprehensive search was made of internet resources in Australia to find current diagnostic and genetic information on Down syndrome. The search terms were 'Down syndrome', 'genetics', and 'diagnosis'. A number of sites were searched, although the key sites used were Baby Center Australia, Pregnancy Birth & Baby, and Your Genome. Other resources such as nursing textbooks were also used to gather research material to ensure a comprehensive approach. The nursing textbooks used for this review were 'Maternal, fetal, & neonatal physiology 4ed.' by Blackburn (2013), and 'Merenstein & Gardener's handbook of neonatal intensive care 8ed.' by Gardner et al. (2016).

An integrative review can play an important role in evidence-based nursing by detailing specific understandings of healthcare issues and practices. This type of review is comprehensive which summarises past empirical and theoretical literature, it surveys a diverse set of data on a particular healthcare issue, in this case; Down syndrome (Whittemore and Knafl, 2005).

3. The genetics associated with down syndrome

During the prenatal period (conception to birth), pregnant women will experience major physiologic and psychologic changes that support maternal adaptations, support foetal growth and development, and preparation for birth. Supporting this development are the placenta, foetal membranes (amnion and chorion), and amniotic fluid. These structures protect and nourish the embryo and foetus and are essential for survival, growth, and development (Blackburn, 2013). Human development begins with the fertilisation of an ovum (female gamete) by a spermatozoon (male gamete). When a spermatozoon comes into contact with the ovum, the zona pellucida and the plasma membrane fuse, preventing entry by other sperm (Kenner and Lott, 2014). During fertilisation, meiotic cell division occurs where the male and the female pronucleus fuse. This results in two haploid numbers (23) of chromosomes (22 autosomes and 1 sex chromosome) from each gamete cell. The zygote is formed and contains the diploid number (46) of chromosomes necessary to create a unique human being (Blackburn, 2013).

Genetic disorders can influence the course of the foetal development and pregnancy and have implications for both the mother and the baby (Blackburn, 2013). All human cells, except for the gamete cells (ovum and sperm), normally contain 46 chromosomes (diploid number) consisting of 1 pair of sex chromosomes and 22 pairs of autosomes (Blackburn, 2013). Chromosomes are made up of genes that determine how the foetus forms in utero and how the baby grows after birth. They also influence physical characteristics, such as eye and hair colour, and the probability of developing a disease in the future (Perkins, 2017).

The genetic basis of Down syndrome was first described by Jerome Lejeune in 1959 (Sheets et al., 2011). An individual with Down syndrome has an additional copy of chromosome 21 (HSA21), either full or partial. The almost complete sequence of the long (q) arm of HSA21 was published in 2000, however the complete sequence of the short (p) arm has not been completed

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