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Cardiac disease in Down Syndrome: literature review and international expert consensus in collaboration with Down Syndrome International (DSi)

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Abstract

Background: Congenital heart disease is common in patients with Down syndrome, yet clinical recommendations relating to its diagnosis and management in this patient group are lacking.

Main body: We discuss the ongoing collaboration between an international panel of cardiovascular experts and expert stakeholders from Down Syndrome International, an international disabled people's organisation with membership of organisations and individuals from 136 countries worldwide. The aim of this collaboration is to describe best clinical practice, focusing on 10 key areas relating to Down syndrome and cardiac disease, from prenatal diagnosis to the care of patients in areas of differing resource availability.

Conclusions: The planned expert consensus statement on cardiac disease in people with Down syndrome aims to foster communication between experts, direct future research and inform future practice guidelines for the diagnosis and management of cardiovascular disease in people with Down syndrome.

Keywords: Down syndrome, Trisomy 21, Congenital heart disease, Epidemiology, Prenatal diagnosis, Cardiac surgery, Long-term outcomes, Healthcare resources

Introduction

Cardiovascular disease is one of the most common health problems in people with Down syndrome. Congenital heart disease (CHD) of various types complicates up to 50%, and there is a predisposition to the development of pulmonary hypertension and other acquired cardiovascular conditions as a result of related co-morbidities such as obesity and sleep apnoea.[1, 2] Even though the impact of cardiovascular conditions on the long-term outcome of people with Down syndrome is well-described,[3, 4] current Down syndrome-specific recommendations focus mainly on non-cardiovascular health. When cardiac disease is considered in clinical guidelines, the focus is on acquired cardiovascular conditions, e.g. lipid management, coronary artery and cerebrovascular disease, rather than on CHD.[5, 6] Furthermore, guidance for specialists who provide CHD-specific care to people with Down syndrome is lacking. Over recent decades, for better or worse, CHD management in people with Down syndrome has differed significantly to that of the general population, and between different countries and healthcare systems, and is a topic often neglected by the wider medical community. The

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goal of the proposed project is to develop an evidence-based expert consensus document for the diagnosis and management of CHD for children and adults with Down syndrome, which in the future may lead to the development of detailed practice guidelines in this area.

Main body

Advocacy for people with Down syndrome around the world is the focus of the work of Down Syndrome International (DSi), which includes the training and education of healthcare providers and of the Down syndrome community. DSi is an international disabled people's organisation with a membership of organisations and individuals from 136 countries worldwide, committed to improving the quality of life for people with Down syndrome.[7] Cardiovascular experts from around the world have joined forces with DSi to develop a state-of-the-art review and expert consensus statement on cardiac disease in people with Down syndrome. Ten key areas relating to the care of patients with Down syndrome and cardiac disease have been developed using feedback from key stakeholders from the DSi global network and the working group of experts, which will form the basis of the statement:

1. Incidence, present and future, and types of CHD in Down syndrome;
2. Best practice for prenatal and neonatal diagnosis, and specific management needs arising during this period;
3. Optimal timing of repair of CHD in Down syndrome and the risk of developing pulmonary hypertension;
4. Perioperative risks, complications and optimal care;
5. Long-term complications and outcome of CHD in Down syndrome;
6. Optimal follow-up and long-term care for patients with CHD and/or pulmonary hypertension and Down syndrome;
7. Acquired cardiac and non-cardiac comorbid considerations to aid heart disease-related decision-making;
8. The influence of learning disabilities on the practical management of patients with Down syndrome and CHD;
9. The optimal approach to diagnose and manage patients with Down syndrome in areas of different resource availability, including healthcare resources;
10. Future research needs and challenges in Down syndrome research.

Expert consensus statements for each topic will be formed from a formal review and synthesis of the literature. The statements will address current best practice in diagnosis, repair and issues arising during lifelong follow-up, including promoting exercise to maintain cardiovascular health. Supporting text will review available evidence and differences that exist in the provision of care across borders and in different healthcare settings. The project will inform consultations and guide diagnosis, treatment and the long-term follow-up of people with Down syndrome affected by congenital heart and other cardiovascular diseases.

Conclusions

Through this work, we also aim to improve communication between experts across borders, stimulate discussion and the creation of training resources, direct future research and, ultimately, allow the development of international practice guidelines for the diagnosis and management of cardiovascular disease in people with Down syndrome.

Abbreviations

CHD: Congenital heart disease; DSi: Down Syndrome international

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Authors' contributions

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Competing interests
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