International Journal of Integrated Care

Garami, Anna 2020 22q11 Deletion Syndrome: Neonatal Workup and Childhood Follow-up *International Journal of Integrated Care, 21(S1)* : A159, pp. 1-8, DOI: doi.org/10.5334/ijic.ICIC20312

CONFERENCE ABSTRACT

22q11 Deletion Syndrome: Neonatal Workup and Childhood Follow-up

ICIC20 Virtual Conference – September 2020

Anna Garami[1]

1: School Of Medicine, Trinity College Dublin, Dublin, Ireland

Background

22q11 deletion syndrome (also known as DiGeorge syndrome and velocardiofacial syndrome) is the most common microdeletion syndrome. The condition typically presents with one or more of congenital heart disease, palatal and ear nose and throat abnormalities, hypocalcaemia and hypoparathyroidism, immune deficiency and developmental delay. Given the wide variety of medical and surgical manifestations of this syndrome, diagnosis and follow-up if often a clinical challenge.

Objective

The aim of this study was to review current literature and guidelines on 22q11 deletion syndrome to develop a clinical tool summarizing avaible evidence. This is to aid in recognition, investigation and appropriate follow-up from the neonatal period.

Method

A systematic literature search was conducted to gather all available evidence pertinent to the diagnosis long-term care of neonates and children with suspected or confirmed 22q11 deletion syndrome. Furthermore, current published guidelines, consensus statements and expert opinion was included.

Results

The initial search returned 446 results, of which 57 articles were included in the final analysis. The literature reflected the wide phenotypic variation seen 22q11 deletion syndrome. The most common presentations were cardiac malformations, developmental delay, palatal abnormalities, immunodeficiency and hypocalcemia. Follow-up is very much dependent on the specific manifestations, with limited evidence supporting any universal best practice approach. Based on the review conducted by this group, a clinical tool (in the form of a single-page flow-chart) was developed to outline the key areas in the long-term care of patients with 22q11 deletion syndrome.

Conclusion

A multidisciplinary approach to this complex syndrome is essential as the age and features at the time of diagnosis are variable. Neonatal diagnosis is becoming more common, and comprehensive checklists with

evidence-based recommendations are valuable in clinical practice. A clinical tool was designed by this group with the intent of guiding decision making, integrating the many aspects of care in this population, and ultimately promote better long-term outcomes for patients.