

Incidence of an underlying syndromic diagnosis in children with an antenatally-detected, isolated cleft lip with or without cleft palate: a regional analysis

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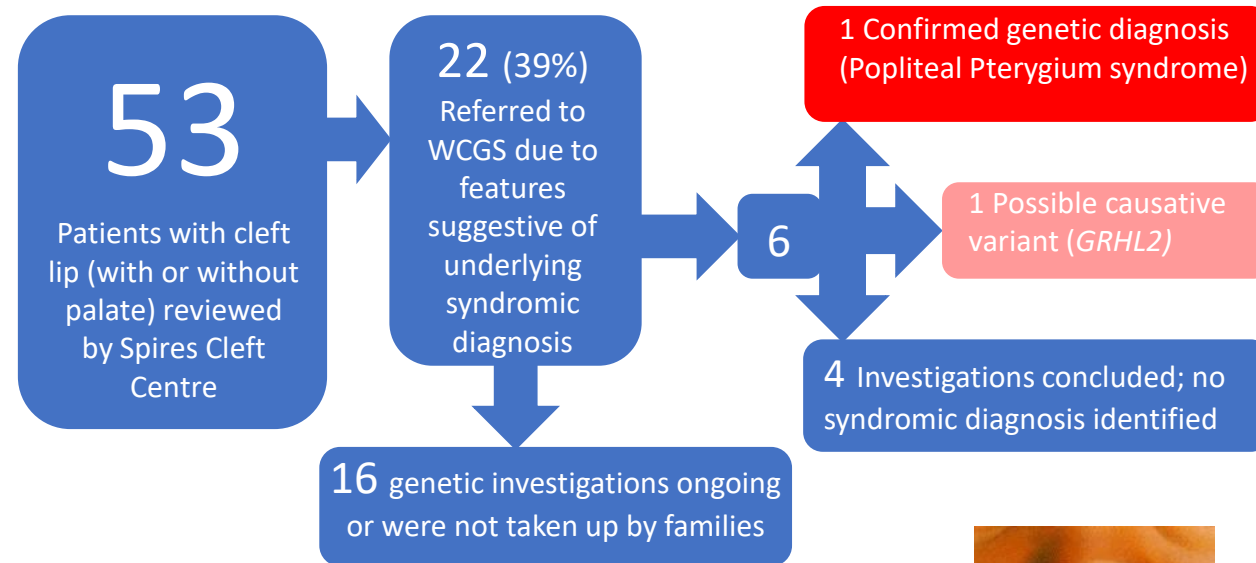
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Background

It is estimated that cleft lip, with or without cleft palate, represents part of a wider syndrome in approximately 15-30% of cases¹. In many of these syndromic cases, additional anomalies will be apparent antenatally. However, there is limited longer term outcome data for apparently isolated cleft lip detected antenatally. It is thought that in this group of patients, the chance of having an underlying syndromic diagnosis is likely to be significantly lower. The aim of this study was to determine the incidence of an underlying genetic or syndromic diagnosis in antenatally-detected isolated cleft lip with or without cleft palate.

Reference

¹ Cleft Lip and Palate Association
<https://www.clapa.com/>



Methods

Data were collected prospectively from the Spires Cleft Centre and Wessex Clinical Genetics Service (WCGS) databases, for a period between June 2020 and December 2022. The records of patients referred to the WCGS were examined to determine whether a genetic diagnosis had been made.

Findings

Thirty-nine percent of babies born with an antenatally detected, apparently isolated cleft lip with or without cleft palate had features postnatally that warranted referral to the Clinical Genetics service. However, in 66.7% (4/6) of those that completed genetic testing, no genetic diagnosis was detected. This study provides useful information for the counselling of couples who have received an antenatal diagnosis of isolated cleft lip with or without cleft palate.

Limitations

We acknowledge that due to the small size of this study, the findings are not amenable to statistical analysis. However, both the results of this study and our experience in counselling couples who have received an antenatal diagnosis of isolated cleft lip, with or without cleft palate, indicate the value of further, more extensive, research in this area.

