

**EVALUATION OF THE CLINICAL FEATURES AND
IDENTIFICATION OF 22q11 DELETION AMONG
SELECTED PATIENTS WITH CLEFT PALATE**

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ABSTRACT

Cleft palate is one of the commonest congenital malformations of the head and neck region. The incidence of isolated cleft palate in the central province of Sri Lanka was 0.19 per 1000 live births. Micro-deletion of chromosome 22q11.2 is one of the commonest syndromic causes of cleft palate. Objective of the study is to describe the socio demographic background, clinical features and prevalence of the 22q11.2 deletion among patients with cleft palate in Sri Lanka. Patients with cleft palate without cleft lip attending the Regional Cleft Centre & Maxillo-Facial Department, Teaching Hospital, Karapitiya were clinically assessed and semi-quantitative PCR was performed to identify the 22q11.2 deletion.

There were 162 subjects (males 41.36%) with an age range of two weeks to forty nine years. Majority had clefts involving the soft palate (n=125/162; 77.16%). There was a statistically significant ($p < 0.05$) preponderance of cleft palate among females and Moor ethnicity. Low birth weight and growth parameters were identified in preschool aged children. Ninety two (56.79%) had other clinical problems including four possible syndromes. The semi-quantitative PCR did not identify any subjects with a 22q11.2 deletion. The low rate of 22q11.2 deletion identified in this sample may be related to selection bias, non-exclusion of other syndromes or technical limitations but may also reflect a low prevalence of this syndrome in this cleft population. Further investigations are required to validate the semi-quantitative PCR method and generalize these findings to the Sri Lankan cleft palate population.

Key words: Cleft Palate, clinical assessment, 22q11.2 deletion, semi-quantitative PCR, growth parameters.