A Descriptive Study of Amelogenesis Imperfecta including Syndromic Forms

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Amelogenesis Imperfecta (AI) is a developmental disorder of genomic origin, associated with abnormal enamel formation. The aim of the present study was to identify different clinical presentations and inheritance patterns observed in Sri Lankan children affected with AI.

Children, who presented to the Division of Paedodontics, Faculty of Dental Sciences, University of Peradeniya over a period of two years from 2008 to 2010, were screened to identify the patients suffering from AI.

The study sample obtained during this period included twenty children with AI, belonging to 16 families with four families each contributing two children with AI. The mean age of the present study sample was 12 years (range: 3-17 years). Female predilection (12/20) with a male to female ratio of 2:3 was also noted. With reference to racial distribution, majority of the affected children were Sinhalese 65% (13/20) followed by 15% (3/20) Tamils and 20% (4/20) Muslims. Out of the twenty patients with AI, 45 % (9/20) had hypoplastic form (Type I) while 30% (6/20), 15% (3/20), 10% (2/20) had hypomaturation (Type II), hypocalcification (Type III), hypoplastic/ hypomaturation form (Type IV) of the disease, respectively. A positive family history could be identified in 50% (8/16) of the families having children with AI. With pedigree plotting, a positive identification of patterns of inheritance as either autosomal dominant or X-linked pattern was possible in eight families. Eight children were considered to show autosomal recessive pattern of inheritance as these children had parents in consanguineous marriages. The remaining three children, without identifiable pattern of inheritance, were considered as sporadic cases of AI. Children who may have Kohlschutter syndrome, Cone-rod dystrophy and Tricho-dento-osseous syndrome were identified, and are currently undergoing further investigations to confirm the diagnoses.

In the absence of previous published studies, our study reports the first descriptive analysis of subtypes of AI in Sri Lankan context and our findings shed light into lesser-known aspects of this complicated group of conditions. This baseline data could be used to improve the awareness among dental surgeons and to highlight the importance of early accurate diagnosis leading to improved quality of life among patients affected by AI.

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