Ring-14 Chromosomal Syndrome: Characterization, Dental Considerations, and Management

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ABSTRACT

Purpose: To investigate the manifestations and challenges faced by subjects suffering from Ring-14 Chromosomal Syndrome, a rare, genetic condition caused by an abnormal ring formation of chromosome (14). The disease was characterized by comparing patients for: phenotypic similarities; anthropometric criteria; physical appearance; craniofacial, dental and occlusal features, and; oral manifestations including caries and periodontal status. Subjects were evaluated for developmental condition and intellectual disability, and other special needs such as epilepsy and autism-related disorder that could impact the dental management of these patients. The information gathered from this study is intended to raise awareness for this extremely rare disease, both to allow health professionals to gain familiarity with the signs and symptoms and to encourage genetic testing early on. With only 50 cases reported worldwide, the disease is largely under-diagnosed and not fully studied, as the dental management of these patients also remains remarkably unaddressed. The investigation provides tips and recommendations that dentists may find helpful to manage r(14) children more effectively, as many are prone to seizures, behavioral outbursts, and unable to speak.

METHODOLOGY

A voluntary survey was administered to the caretakers of 13 r(14) patients who as of 2017 were registered in the (NORD-National Organization for Rare Diseases) global data bank (Ring 14 USA Outreach). The patients were assessed for age, gender, geographic distribution, phenotype, physical appearance, maxillofacial characteristics, presence of oral conditions and abnormalities, malocclusion, epileptic seizures, cognitive abilities, speech, muscle tone, nutrition, autism, and other developmental and behavioral points of interest.

RESULTS

Of the 13 patients queried, 7 were male and 6 were female. The age of the patients ranged from 5 to 49 years of age. Ten patients were of European ancestry and three were Hispanic, all residing across the U.S. The majority of patients were diagnosed as infants, shortly after commencement of uncontrollable seizures. All the patients were microcephalic and presented with Class II malocclusions. More frequent occlusal anomalies and conditions included diastemae of the anterior teeth, congenitally missing teeth, crowding, and drooling. The majority of subjects was unable to speak, suffered from intractable seizures, and frequently exhibited behavioral outbursts.

CONCLUSION

A child with r(14) may present a considerable challenge to the dentist and staff, but the dental problems of r(14) children are for the most part like any other patient and can often be handled by the dentist. Depending on the severity of symptoms, some children with r(14) may be as treatable in the dental office as any other child.