

大学院特別講義のご案内

- ◆ 日時： 2021年8月27日 22:00-23:30
- ◆ 場所：オンライン（参加希望者は矯正科まで）
- ◆ 講師：黒坂 寛

Osaka University Dental Hospital Dept of Orthodontics

- ◆ 演題：The role of orthodontist in the treatment of cases with rare genetic diseases
- ◆ 要旨：Craniofacial deformity comprises approximately 30% of congenital diseases and could cause severe malocclusion. Cleft lip and palate, retrognathia, tooth anomalies are some of the representative phenotype of congenital craniofacial defects. Many rare undiagnosed diseases are also known to associate with variety of craniofacial defects. Our clinic in association with Initiative of Rare and Undiagnosed Disease (IRUD) are investigating genetic cause of rare diseases by performing whole exome sequencing. As a result, we have detected some novel gene mutations in undiagnosed patients and thus diagnosed these patients as pseudo hypoparathyroidism and Baraitser-Winter syndrome. Interestingly there were many unreported craniofacial defects in these patients which indicate the significance of precise description of craniofacial defects in rare diseases to improve the diagnosis rate. We are also producing novel disease models using mice, zebrafish and cell lines to further investigate the molecular and cellular etiology of these rare diseases which possibly could lead into developing new therapeutic methods.



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