Dental management of 49,XXXXY syndrome with taurodontism: A case report

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Abstract
A Japanese male with a karyotype of 49,XXXXY was presented for dental treatments from 6 years to 20 years of age. The patient had pulmonary stenosis as well as mental retardation, hypogonadism, fifth finger and toe clinoctaly, genu valgus, pes cavus and mandibular prognathism. Dental manifestations revealed multiple caries and taurodontism of both primary and permanent dentitions and hypodontia in permanent dentition. Dental management of a patient with 49,XXXXY syndrome is important to accomplish both controlling behavioral problems and medical considerations for preventing infective endocarditis.

Case Report
In April 1994, a 6-year and 10-month-old Japanese boy, with health problems, was referred to the pediatric dental clinic at the Tsurumi University Dental Hospital presenting dental pain associated with the mandibular primary molars. The patient was born at 37 weeks gestation by Cesarean section. At birth, his weight was 2,090 g and his height was 43.5 cm. He had been indicated with a karyotype of 49 chromosomes and X-chromosomal aneuploidy and pulmonary stenosis in the examination at birth. He began walking late at 3 years of age, and it became obvious that physical and intellectual development was also late and deficient.

At first visit, he was not able to speak to a dental staff, but seemed to catch their speech. Physical examination revealed bilateral fifth finger and toe clinoctaly and his gait was unsteady. Clinical and radiographic examination of the dentition revealed extensive caries, especially deep carious lesions of the mandibular primary molars that cause his complaint of occasional dental pain. Periapical radiographs revealed taurodontism in the mandibular primary molars, and the maxillary molars showed same features (Fig. 1). His upper and lower primary
incisors was in cross-bite relationship.

Dental treatments were performed under prophylactic antibiotic administration according to the American Heart Association guidelines to prevent infective endocarditis. Access cavity preparations of the lower primary molars were performed under local anesthesia by using tell, show, do method (TSD method). The patient seemed relatively cooperative.

In compliance with the wishes of the patient’s mother and with his consent, a 10 mL blood sample was drawn from vein for karyotyping. All of 20 cells counted had 49,XXXXY chromosomes (Fig. 2).

Second visit was at 12 years and 11 months of age with a complaint of decayed teeth. His height was 133 cm and body weight was 29.5 kg (standard of the same age in Japanese boy: 155.6 cm and 44.1 kg, respectively). He could talk with the dental staffs. Facial appearances showed orbital hypertelorism (4.0 cm), epicanthal fold, internal strabismus, saddle nose, bilateral abnormality of auricles and prognathism, but no upslanting palpebral fissures (Fig. 3).

Clinical and panoramic radiographic examination of the dentition revealed multiple caries, shovel-shaped upper central incisors, small peg lateral incisors, transposition between upper left canine and bicuspids, and taurodontic teeth (permanent upper both first molars and second molars, permanent lower left first molar and second molar, permanent lower right first molar), and agenesis of permanent lower left second premolar and permanent lower right second molar (Fig. 4).

Composite resin restorations, chromium-nickel crowns, endodontic treatments, tooth extractions and a dental prophylaxis were provided.

The patient was cooperative and tolerated dental treatments well, but endodontic procedures of taurodont teeth were challenging technically because the teeth had large pulp chambers and their furcations positioned near apical regions.

Cephalometric analysis revealed a skeletal mesio-occlusion due to a long corpus mandibula and a forward rotation of the mandible (Fig. 5). Orthodontic treatment did not performed, because the patient refused it.

Last visit was at 18 years and 1 month of age with a complaint of tooth fracture. Three primary teeth (primary upper right canine, primary lower both second molars) have been retained and 23
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permanent teeth existed in oral cavity. By visual and periapical radiographic examination, the fractured tooth (permanent upper left canine) was extracted as conservative treatment was impossible. We lasted this patient’s dental management to 20 years of age (Fig. 6).

Classification on the degree of taurodontism
A periapical radiograph of the affected tooth was

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Fig. 4  Panorama X-ray photo at 12 years and 11 months of age. ( ); unerupted.

Fig. 5  Facial diagram at 12 years and 11 months of age. (Dotted line: this case, Solid line: standard)

Fig. 6  Panorama X-ray photo at 20 years of age. Endodontic treatments in taurodont teeth succeeded in general.

Fig. 7  Diagram shows method of measuring the two variables (Y, Z).
Y is the vertical height of the chamber (distance from lowest part of roof to highest level of floor of pulp chamber), and Z is the distance from the lowest part of roof of pulp chamber to the apex of longest root. Taurodontism Index (TI) = Y/Z × 100.
By calculating TI, the degree of taurodontism is classified in three types (25.0–49.9: hypo-, 50.0–74.9: meso-, 75.0–100.0: hyper-taurodontism)
traced and the two variables on the tracing film were measured as showing a diagram (Fig. 7). And the taurodontism index (TI) was calculated by the formula reported by Keene\textsuperscript{11}. The degree of taurodontism by TI was classified three types: hypo-, meso- or hyper-taurodontism.

The results of measurements of primary and permanent molars show in Table 1.

### Discussion

The majority of case reports cases have not distinguished the 49,XXXXY syndrome from Klinefelter syndrome (47,XXY), and these patients are frequently labelled as having Klinefelter syndrome or as being a “Klinefelter variant”. However, today, it has been recognized that the 49,XXXXY syndrome has its own phenotype, and genotype and is not Klinefelter syndrome, as Peet \textit{et al.}\textsuperscript{4} delineated the 49,XXXXY syndrome was different from Klinefelter syndrome because of its distinct clinical features, especially the prevalence of congenital heart defects.

In the present case, the patient had pulmonary stenosis as well as mental retardation, hypogonadism, short stature, genu valgus, muscular hypotonia, fifth finger and toe clinodactyly, orbital hypertelorism, saddle nose, and abnormality of auricles, but no radioulnar synostosis.

Facial appearance revealed a coarse face with mandibular prognathism, but without eyes slanting upward as found in Down’s syndrome that Gorlin \textit{et al.}\textsuperscript{3} had described.

Dental findings revealed multiple taurodontic teeth as reported in several cases\textsuperscript{3–5,8,9} with 49,XXXXY syndrome. Some authors have described the relationship between taurodontism and an extra X chromosome\textsuperscript{5,12,13}. Jaspers and Witkop\textsuperscript{5} stated that there was no simple association of the degree of taurodontism and the number of X chromosomes, but in general, patients with more severe forms of the trait — meso- or hyper-taurodontism — were more likely to have X-chromosome aneuploidy. Varrela \textit{et al.}\textsuperscript{13} indicated that there was positive correlation between the degree of taurodontism and the number of X chromosomes. In the present case, primary and permanent molars revealed hypo-, meso- or hyper-taurodontism except primary upper first molars.

Other features were shovel-shaped upper central incisors, as Gardner \textit{et al.}\textsuperscript{12} described same manifestations, and congenital absence of permanent tooth germs as described in few cases\textsuperscript{7,9}. Transposition between upper left canine and bicuspid was first original finding that had never reported.

Endodontic treatment of taurodont tooth was challenging, because it requires special care in handling and identifying the orifices of root canals. Some authors have described successful completion in endodontic treatment of taurodont tooth\textsuperscript{13–15}. We also succeeded its treatments, but might manage more easily if could have complete caries treatments in more early.

### Conclusion

Dental management of a patient with 49,XXXXY syndrome is important to accomplish both controlling behavior problems and medical considerations for preventing infective endocarditis as well as to maintain good oral hygiene.

### Acknowledgments

Written consent was obtained from the patient and his mother for publication of this case report.
References


