Endodontic management of a hypertaurodontic tooth associated with 48, XXYY syndrome: A review and case report

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Abstract

Taurodontism is a developmental anomaly of a tooth characterized by large pulp chamber and short roots. Patients with multiple taurodontic teeth are associated with the probability of a systemic syndrome or chromosomal anomaly. This is the first reported incidence of the endodontic management of a hyper taurodontic mandibular second molar in a patient diagnosed with 48, XXYY syndrome.

Keywords: XXY/Klinefelter syndrome; 48, XXYY syndrome; hypertaurodontism

INTRODUCTION

XXYY syndrome also known as 48, XXYY syndrome or “double male syndrome” is a rare sex chromosome aneuploidy condition characterized by presence of two extra X and Y chromosomes with an incidence of 1:18,000-1:40,000 male births.[1-3] This syndrome is clinically manifested later in life with developmental delays, learning disabilities, behavioral problems, and delayed or incomplete puberty. Many cases go undiagnosed as most of the above mentioned abnormalities do not develop until early puberty.[4,5]

The term taurodontism was coined by Sir Arthur Kein to describe the bull-like appearance of the teeth.[6] Taurodontism can be defined as a morphological variation of a tooth in which the pulp chamber is vertically elongated and the roots are reduced in size. Taurodontic teeth have large pulp chambers with apically positioned furcation area.[7] The etiology of taurodontism is attributed to the failure of Hertwig epithelial root sheath diaphragm to invaginate at the proper horizontal level.[6] Based on the severity, taurodontic teeth can be classified as hyp-, meso-, and hypertaurodont forms. Hypotaurodont expresses the least pronounced changes, while mesotaurodont expresses moderate variation, and hypertaurodont is characterized by the most severe variation in which the roots bifurcate or trifurcate close to the apices.[8]

Multiple taurodontic teeth may occur as one of the dental manifestations of XXYY syndrome. The presence of taurodontic teeth can facilitate early recognition of this disorder. This could provide means for an effective systemic rehabilitation.[4] This is the first reported incidence of the endodontic management of a hypertaurodontic mandibular molar tooth in a patient diagnosed with 48, XXYY syndrome.

CASE REPORT

A 19-year-old male reported to the postgraduate department of endodontics of our dental school, for management of multiple carious teeth. During history taking, it was elicited that the patient was a school dropout due to difficulty in coping with the educational curriculum. It was also evident, that the patient had below average communication skills with low self-esteem. The patient’s medical history was noncontributory. The patient’s dental history revealed episodes of recurrent caries and multiple extractions. Extraoral examination revealed retruded mid face with mandibular prognathism [Figure 1].

Intraoral examination revealed dental caries in relation to teeth 15, 37, and 47 and multiple missing teeth (14, 13, 22, 23, and 36). An orthopantomograph revealed multiple taurodontic teeth in relation to 37, 46, 47, 17, 26, and 27 [Figure 2a]. The taurodontic teeth exhibited large pulp

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chamber extending beyond the cervical area and reaching the furcation suggestive of hypertaurodontism. On clinical examination, tooth 37 showed no response to electric and cold test. The tooth was nonsensitive to percussion, and radiographic interpretation was suggestive of asymptomatic apical periodontitis. A decision was made to perform orthograde root canal treatment in 37. The patient was informed about the complex root anatomy and possible complications. Patient consent was obtained prior to commencement of treatment.

The complete endodontic management of the case was performed by one of the authors, while the other author took care of post-endodontic restoration and clinical follow-up with department of oral surgery. The patient was anesthetized with 1.7 ml of 4% articaine with 1:100,000 epinephrine (Septocaine, Septodont, New Castle, DE, USA) and access opening was performed under rubber dam isolation. Endodontic treatment in taurodontic teeth has been described as difficult as the complex morphology could hamper the location of the root canal orifices. The access opening and canal tracing was performed with the help of adental operating microscope (G6, Global Microscope, USA). Careful inspection and exploration revealed a large coronal pulp chamber extending 8-10 mm below the cervical margin (cementoenamel junction (CEJ)). The presence of two canals (mesial and distal) bifurcating 3-4 mm short of the radiographic apex was identified. An electronic apex locator (Root ZX, Morita, Tokyo Japan) was used to determine the working length. The shaping and cleaning of all canals was done using nickel-titanium rotary instruments, K3 (Sybron Endo, Orange, CA, USA). Irrigation was done with 5.2% sodium hypochlorite solution (NaOCl) during shaping and cleaning of the root canal system. Passive ultrasonic irrigation (PUI) activation was employed using # 20/0.00 taper SS noncutting ultrasonic tip (Irrisafe, Satelec, Acteon, Merignae, France) followed by 1 min flush of 17% ethylenediaminetetraacetic acid (EDTA; Smearclear, SybronEndo, CA, USA). Final irrigation was done using 5.2%NaOCl. The canals were dried using sterile paper points and dressed with intracanal calcium hydroxide medicament and the tooth was sealed temporarily with an intermediate restorative material (CavitG, 3M-ESPE, Seefeld, Germany).

The patient was asked to return after 2 weeks. At the second appointment, copious irrigation was done in order to remove the calcium hydroxide and the canals were made dry with sterile paper points. Following radiographic control of the fit of gutta-percha mastercone (Guttapercha, SybronEndo, CA, USA), the apical part of the main root canals was obturated with vertically compacted warm gutta-percha using the Touch&Heat 5004 device (Sybron Endo, CA, USA). The coronal portion of the main canal was backfilled with thermoplasticized gutta-percha using the Obtura Ilgun (Obtura-Spartan Corp, Fenton, MO, USA). The access cavity was sealed with dualcuredcomposite resin (ParaCore, Coltene/Whaledent, NJ, USA). A full coverage crown was restored on the tooth after 3 weeks’ time. The patient was then referred to the Department of Oral Surgery for surgical correction of prognathic mandible. The surgical intervention was performed in two stages. The first stage involved a bilateral sagittal split osteotomy followed after 3 months by a second stage surgical advancement genioplasty procedure. Follow-up was at 6-months and 2-year intervals. Patient remained asymptomatic at these times.

**DISCUSSION**

A normal individual has 22 pairs of autosomes (numbered chromosomes) and one pair of autosomes and a single X and Y chromosome, making a karyotype or chromosomal makeup of 46, XY. Similarly, a normal female has 22 pairs of autosomes and two X chromosomes, making a karyotype 46, XX.

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**Figure 1:** (a) Preoperative extraoral photograph showing mandibular prognathism. (b and c) Postsurgical photograph after bilateral sagittal split osteotomy and genioplasty

**Figure 2:** (a) Preoperative orthopantomograph before the root canal treatment. (b) Immediate postoperative radiograph. (c) A 2-year follow-up post endodontic restoration in 37
48, XXYY syndrome is characterized by an extra X chromosome and an extra Y chromosome. This, in addition to the 22 pairs of autosomes, males with 48, XXXY syndrome have four sex chromosomes, making a karyotype 48, XXYY. Successive meiotic nondisjunctual events during spermatogenesis, resulting in a XYY sperm have been attributed as the most probable etiological mechanism of 48, XXYY syndrome. The presence of extra sex chromosomes may lead to altered deoxyribonucleic acid (DNA) methylation at various loci in genome leading to altered gene expression and subsequent phenotype variation.\(^\text{[2,3]}\)

According to Gardner and Girgis,\(^\text{[9]}\) a patient with multiple taurodontic teeth without known association of any systemic syndrome should be consulted for chromosomal analysis, as there is a higher association of taurodontic teeth with X-chromosome aneuploidy syndromes. Hence, we referred our patient to the department of human genetics for chromosomal analysis. Chromosome analysis with GTG banding with a resolution of 450-550 bp, showed a numerical abnormality in all cells analyzed, with the presence of an extra X and an extra Y chromosome resulting in 48 chromosomes and a diagnosis of 48, XXYY syndrome was made [Figure 3].

The prevalence of taurodontism ranges from 2.5 to 11.3% of the human population.\(^\text{[10]}\) Taurodontism can occur either as an isolated manifestation or in association with various systemic syndromes. A literature search for relevant articles regarding endodontic management of taurodontic tooth associated with systemic syndromes was performed using Ovid MEDLINE®, Cochrane Database of Systemic Reviews, Embase, and PubMed. Table 1 summarizes the chronological order of case reports of taurodontism associated with systemic syndromes.

Table 1: Chronological reports of systemic syndromes associated with taurodontism

<table>
<thead>
<tr>
<th>Year</th>
<th>Syndrome</th>
<th>Author</th>
<th>Chromosomal disorder</th>
</tr>
</thead>
<tbody>
<tr>
<td>1970</td>
<td>Mohr syndrome</td>
<td>Gorlin(^\text{[11]})</td>
<td>Autosomal recessive</td>
</tr>
<tr>
<td>1973</td>
<td>Klinefelter syndrome</td>
<td>Mednick(^\text{12})</td>
<td>Additional X chromosome</td>
</tr>
<tr>
<td>1978</td>
<td>Tricho-onycho-dental syndrome</td>
<td>Koshiba \text{et al}(^\text{13})</td>
<td>Autosomal dominant</td>
</tr>
<tr>
<td>1981</td>
<td>Downs syndrome</td>
<td>Jaspers(^\text{14})</td>
<td>Additional 21 chromosome</td>
</tr>
<tr>
<td>1991</td>
<td>Prader- Labhart-Willi syndrome</td>
<td>Bassarelli \text{et al}(^\text{15})</td>
<td>Gene deletion at chromosome 15</td>
</tr>
<tr>
<td>1997</td>
<td>Lowe syndrome</td>
<td>Tsai and O’Donnell(^\text{16})</td>
<td>X-linked recessive</td>
</tr>
<tr>
<td>1998</td>
<td>Tricho-dento-osseous syndrome</td>
<td>Spangler \text{et al}(^\text{17})</td>
<td>Autosomal dominant</td>
</tr>
<tr>
<td>1998</td>
<td>Wolf-Hirschhorn syndrome</td>
<td>Breen(^\text{18})</td>
<td>Partial deletion of the terminal portion of the short arm of chromosome 4</td>
</tr>
<tr>
<td>1998</td>
<td>Ellis-van Creveld syndrome</td>
<td>Hunter and Roberts(^\text{19})</td>
<td>Autosomal dominant</td>
</tr>
<tr>
<td>2001</td>
<td>Apert syndrome</td>
<td>Terezhalmy \text{et al}(^\text{20})</td>
<td>Autosomal dominant</td>
</tr>
<tr>
<td>2002</td>
<td>Seckel syndrome</td>
<td>Seymen \text{et al}(^\text{22})</td>
<td>Autosomal recessive</td>
</tr>
<tr>
<td>2003</td>
<td>McCune-Albright syndrome</td>
<td>Akintoye \text{et al}(^\text{22})</td>
<td>Autosomal dominant</td>
</tr>
<tr>
<td>2003</td>
<td>Lenz microphthalmia syndrome</td>
<td>Erns \text{et al}(^\text{23})</td>
<td>X-linked recessive</td>
</tr>
<tr>
<td>2003</td>
<td>Kabuki syndrome</td>
<td>Petzdold \text{et al}(^\text{24})</td>
<td>Autosomal dominant</td>
</tr>
<tr>
<td>2005</td>
<td>Williams syndrome</td>
<td>Axelsson(^\text{25})</td>
<td>Gene deletion at chromosome 7</td>
</tr>
<tr>
<td>2006</td>
<td>Smith-Magenis syndrome</td>
<td>Tomona \text{et al}(^\text{26})</td>
<td>Gene deletion at chromosome 7</td>
</tr>
<tr>
<td>2015</td>
<td>XXXY syndrome</td>
<td>Our present case report</td>
<td>Additional X and Y chromosome</td>
</tr>
</tbody>
</table>

Table 2: Comparison of 47, XXY Klinefelter syndrome and 48, XXYY syndrome (Adapted from Tartaglia \text{et al}., 48, XXXY, 48, XXXY, and 49, XXXXY syndromes: Not just variants of Klinefelter syndrome, Acta Paediatr: 2011; 100(6):851-860)

<table>
<thead>
<tr>
<th>Characteristic features</th>
<th>47, XXY</th>
<th>48, XXYY</th>
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</thead>
<tbody>
<tr>
<td>Parent of origin of extra chromosomes</td>
<td>50% maternal and 50% paternal</td>
<td>100% paternal</td>
</tr>
<tr>
<td>Prevalence</td>
<td>1:650-1:1,000 males</td>
<td>1:18,000-1:40,000 males</td>
</tr>
<tr>
<td>Congenital malformations</td>
<td>Cliniocacytly and common congenital</td>
<td>Cliniocacytly (70%) and congenital</td>
</tr>
<tr>
<td></td>
<td>malformations 18% (inguinal hernia,</td>
<td>malformations 56% (inguinal hernia,</td>
</tr>
<tr>
<td></td>
<td>cleft palate - occult, submucous)</td>
<td>cardiac, radioular synostosis,</td>
</tr>
<tr>
<td>Developmental and cognitive</td>
<td>Speech and motor delays (40-75%);</td>
<td>Speech and motor delays (75-92%);</td>
</tr>
<tr>
<td></td>
<td>50-75% learning disabilities</td>
<td>100% learning disabilities</td>
</tr>
</tbody>
</table>
This case showing 48, XXYY karyotype is recognized as a distinct clinical and genetic entity. This syndrome was previously considered to be a variant of Klinefelter syndrome (47, XXY); however, it is now considered to be a distinct phenotype with more significant cognitive and psychological impairments. The characteristic differences between these two syndromes have been well documented by Tartaglia et al.,[27] and are highlighted in Table 2.

CONCLUSION

Knowledge regarding 48, XXYY syndrome and its dentofacial manifestations is important for early diagnosis and long-term predictable medical and dental health management. An early recognition of such a disorder is crucial in improving the psychological as well a dental quality of life in affected patients.

REFERENCES


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