

Satoyoshi Syndrome with Progressive Orofacial Manifestations: A Case History Report

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A young female patient suffering from Satoyoshi syndrome had the first characteristic signs and symptoms of hair loss and progressive spontaneous intermittent painful spasms of limb muscles at age 6.5 years. Thereafter, she developed chronic diarrhea, amenorrhea, and skeletal deformities. In the orofacial region, she suffered from painful spasms of the masseter (jaw closing) muscles, progressive tooth loss, and degeneration of the mandibular condyles. Treatment with steroids and provision of complete dentures improved the signs and symptoms. Early diagnosis and timely provision of multidisciplinary care can minimize complications in these patients and improve their orofacial functions and quality of life. *Int J Prosthodont* 2017;30:163–167. doi: 10.11607/ijp.4905

Satoyoshi syndrome (also referred to as Komura-Guerri [cramp in the calf] syndrome) is a rare multisystem disorder first reported by Satoyoshi and Yamada in Japan in 1957.¹ It has also been reported in other countries, and about 60 cases have been reported to date.^{1–10} The syndrome is characterized by progressive partial or complete hair loss (alopecia) and painful intermittent muscle spasms that start in the limbs and spread to other muscles. Other signs may include diarrhea, amenorrhea, and skeletal abnormalities such as short stature and deformed joints.^{1–10} The first signs and symptoms can appear as early as childhood or at adulthood (mean: 11 years) and are approximately three times more common in females than in males. The etiology is still unclear but has often been associated with autoimmune diseases.^{4,6} Very limited information is available regarding orofacial-related manifestations of the syndrome.^{2,7,8} The present case history report describes a 26-year-old Chinese female, who in addition to the

commonly reported manifestations of the Satoyoshi syndrome had abnormal orofacial manifestations including progressive painful spasms of the masseter (jaw closing) muscles, tooth loss, and degeneration of the mandibular condyles.

Case History Report

The first characteristic signs and symptoms of Satoyoshi syndrome in the present patient appeared at the age of 6.5 years and included thinning of the hair on the scalp, eyebrows, and eyelashes and spontaneous painful intermittent muscle spasms of the lower limbs that might have occurred several times throughout the day and night. Over-the-counter analgesics were used to control the pain. In the years that followed, the muscle spasms gradually intensified in magnitude, frequency, and duration (lasting seconds to minutes and occurring 10 to 20 times per day) and involved several muscles, including the jaw-closing masseter muscles. She also developed chronic diarrhea. At 9 years of age, she was admitted to the Endocrine Department at Peking Union Medical College Hospital (PUMCH), and at age 15 she was diagnosed with Satoyoshi syndrome based on the above manifestations as well as elevated levels of creatine kinase in the serum (a marker of muscle damage), absence of secondary sex characteristics and menstruation, and several skeletal abnormalities that included short stature and deformed limb joints. Family history was unremarkable. She was treated with steroids (prednisone, 40 mg qd po), vitamin C (200 mg tid po), and an anticonvulsant (carbamazepine, 100 mg bid po). Following 2 months of treatment, there was a significant decrease in the frequency and intensity of muscle spasms (1 to 2 noticeable jaw muscle spasms

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Fig 1 Extraoral and intraoral views of the patient. **(a)** Frontal view showing thin hair on the scalp and eyebrows (partial alopecia) at 16 years of age. **(b)** Intraoral view at age 16 showing missing mandibular first and second molar teeth bilaterally, partially erupted third molars, plaque accumulation and gingivitis on remaining teeth, and patches of depapillation on the tongue. **(c)** Intraoral view at age 16 of edentulous maxilla (except for partially erupted left third molar) and a severely resorbed alveolar ridge. **(d)** Frontal view at 25 years showing regrowth of hair on the scalp and eyebrows. **(e)** Intraoral view at age 25 of the edentulous mandible. **(f)** Intraoral view at age 25 of the edentulous maxilla.

per day), pain intensity (no need for analgesics), and diarrhea. Following 6 months of treatment, the hair on the scalp and eyebrows regrew but remained thin. By age 25 she had a full head of hair and nice eyebrows (Fig 1) and had begun to menstruate regularly.

Orofacial Manifestations

Progressive Tooth Loss

Examination of past orofacial radiographs revealed that at age 9, all tooth crowns except for second and third molars were well erupted into the oral cavity (Fig 2a), compared with an average age of 11.5 years for girls.¹¹ Generally, root formation and alveolar bone growth depend on a normal tooth eruption process. Any deviation from this process may affect the dimension and form of roots and bone.^{12,13} Indeed, the roots of the present patient, in particular those of the molars and canines, were very short and with unfavorable crown-to-root ratios (1:1–3:1). By age 16, all maxillary teeth and the mandibular first and second molars had been lost (Figs 1b, 1c, and 2b) and although the roots of the remaining mandibular anterior teeth, canines, and premolars appeared fully developed, the apical third of the roots was thin and tapered and the third molars had no roots (Fig 2b). Clinical records revealed that the teeth were vital and asymptomatic, with pocket depths of 2 to 4 mm and tooth mobility ranging between Miller Class I and II. The first dental treatments provided at age 16 included oral hygiene instructions, tooth cleaning, and provision of a complete maxillary denture, but no replacement treatment was provided for the missing mandibular molars.

Between 16 and 26 years, the patient had irregular dental appointments, and the maxillary denture was replaced four times due to inadequate stability and retention. Radiographs of the mandibular teeth taken during these years revealed progressive horizontal bone loss and external root resorption in the remaining teeth, which manifested as shortening and rounding of the root apices and widening of the periodontal ligaments (Fig 2c). By age 25 she was completely edentulous (Figs 1e and 1f). One other case history report of Satoyoshi syndrome in a Japanese male patient reports complete loss of teeth by age 17 due to periodontitis.⁷ It is possible that the dental findings of premature root resorption with a widening of the periodontal ligaments, alveolar bone resorption, and early tooth exfoliation are manifestations of the syndrome as in other autoimmune diseases (eg, scleroderma, type 1 diabetes, desquamative gingivitis).¹⁴ However, it is also possible, at least in part, that the dental findings are the result of excessive occlusal forces produced by the repeated spasms of the masseter muscles and subsequent premature root resorption of the primary and permanent teeth,^{15,16} along with irregular dental care, poor oral hygiene, and periodontitis. Lack of prosthetic replacement of missing posterior teeth and provision of adequate occlusal support may have also been a contributing factor. It is also unclear whether endodontic intervention could have prevented the progression of root resorption. For example, endodontic treatment and application of calcium hydroxide for several months could have successfully stopped the external root resorption of a tooth subjected to excessive occlusal forces¹⁶ and of multiple teeth with an idiopathic etiology of external root resorption.¹⁷

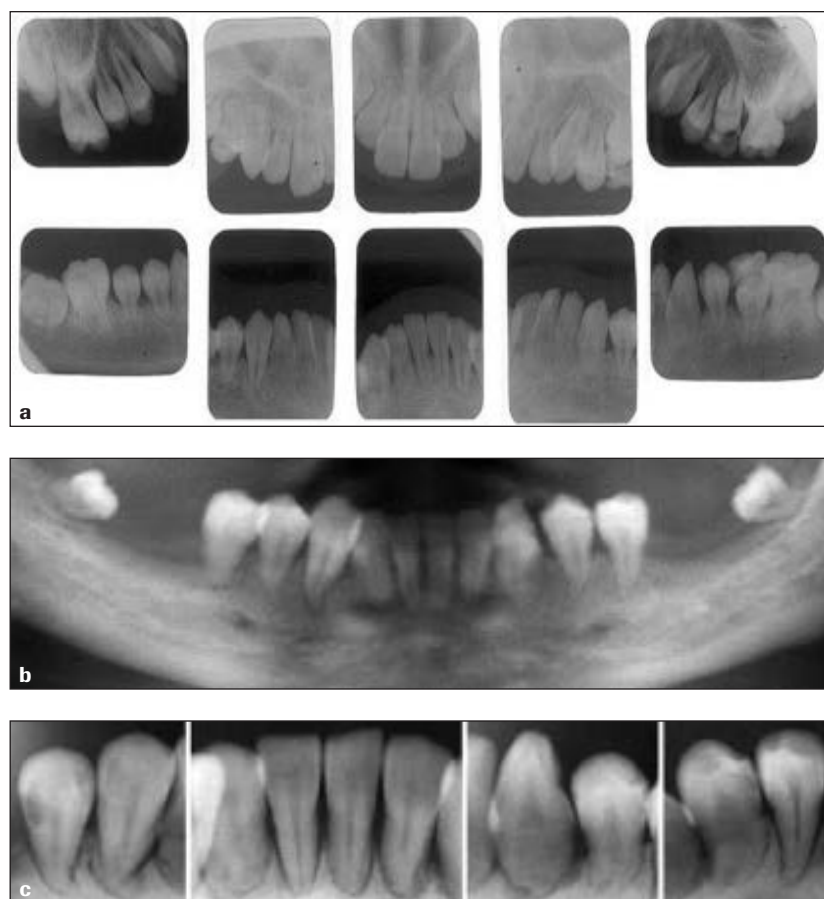


Fig 2 (a) Full-mouth periapical series taken at age 9. Note the normal number of permanent teeth and short roots with opened apical foraminae. (b) Mandibular portion of a panoramic radiograph taken at age 16. Note the missing first and second mandibular molars; short, thin, tapered roots in the remaining teeth; and reduced alveolar bone height around remaining premolars. (c) Periapical radiographs taken at age 21. Note the very short roots with a rounded tip, widened periodontal ligament, and reduced vertical bone support.

At age 26, the patient was provided with a new set of maxillary and mandibular complete dentures with cusplless teeth and balanced occlusions in an attempt to minimize lateral stresses on residual ridges and improve denture stability.¹⁸ Postinsertion evaluation revealed that despite occasional spontaneous involuntary spasms of the masseter muscles along with a right lateral shift of the mandible, the patient adapted well to the new dentures and had no functional concerns. She had gained weight, successfully graduated from a vocational school, and was employed at a local radio station as a broadcaster.

Spontaneous Spasms of the Masseter Muscles

From age 9, the patient had suffered from painful muscle spasms of the masseter muscles, in particular the right. These muscle spasms could occur at any time of day and were accompanied by an involuntary

uncontrolled lateral shift of the jaw to the right. These spasms could last from a few seconds to a couple of minutes and could interfere with her ability to appropriately eat and speak.

Surface electromyographic (sEMG) recordings revealed functional abnormalities in the left and right masseter muscles (Fig 3). While a normal muscle at rest has a characteristic isoelectric baseline (ie, electrical silence), in the present patient the EMG recording at rest showed normal baseline EMG activity with occasional abnormal spontaneous EMG activities in the left and right masseter muscles that could last 30 seconds to 2 minutes. In the left masseter, this involuntary EMG activity had firing frequencies (50–70 Hz) and action potential amplitudes (1.8–2.0 mV) similar to those observed during a voluntary maximal muscle contraction (ie, tooth clenching). However, in the right masseter, the firing frequencies (70–80 Hz) and action potential amplitudes (5.5–11.0 mV) were as much as 5 times higher than those observed during voluntary

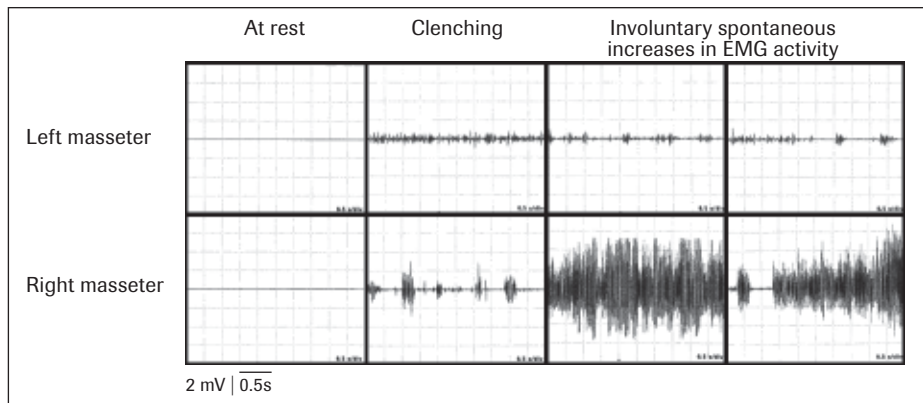


Fig 3 Raw surface-EMG activities recorded in the left and right masseter muscles. At rest, a symmetric isoelectric baseline (ie, no motor unit discharge) could be observed in the left and right muscles. Voluntary maximal muscle contraction (ie, clenching) showed a typical recruitment of motor units in the left masseter muscle and abnormal motor unit recruitment in the right masseter with areas of baseline with or without reduced motor unit activity. At two different time points during involuntary spontaneous increases in EMG activity in the left and right masseter muscles, the motor unit discharge in the right masseter had frequency and action potential amplitudes up to five times higher than those observed during voluntary maximal muscle contraction.

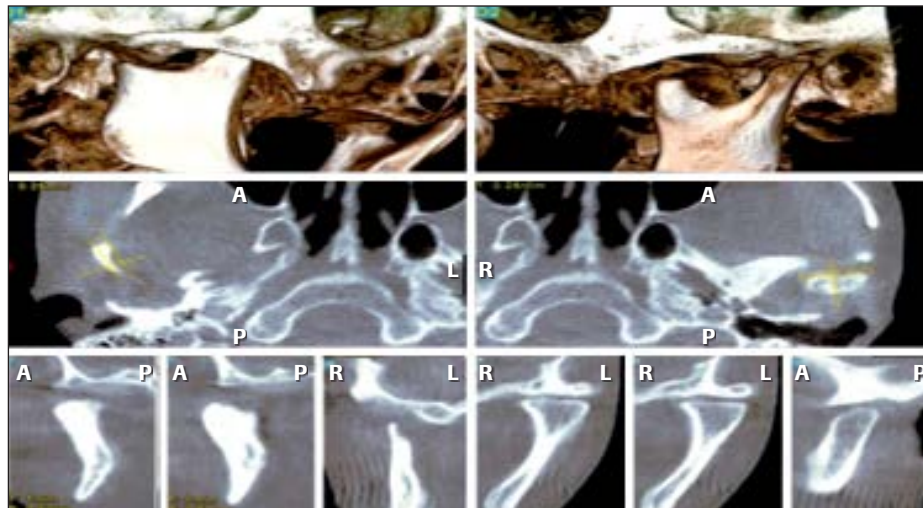


Fig 4 Representative views from a full cone beam computed tomography scan taken at age 26. Note the complete absence of the head and neck of the right condyle and the flattening of the articular surface of the left condyle. A = anterior; P = posterior; L = left; R = right.

maximal muscle contractions. At the time of increased sEMG activity, involuntary and uncontrolled spasms of the masseter muscle and a lateral shift of the jaw could be observed clinically.

The EMG recordings during voluntary maximal tooth clenching (Fig 3) also showed an abnormal pattern of muscle activity. While the EMG recording for the left masseter demonstrated a fairly typical recruitment pattern of motor units and spikes of motor units filled the baseline EMG recordings and became

indistinguishable due to overlaps, the right masseter EMG recording revealed an abnormal recruitment of motor units with areas of baseline-level activity with or without reduced motor unit activity.

Degeneration of the Temporomandibular Joints

Clinical examination at age 26 showed no evidence of periauricular erythema or swelling, no tenderness

to palpation of the temporomandibular joints (TMJ) and masticatory muscles, and no complaints of any orofacial pain. Maximum mouth opening was within normal limits (38 mm), but with a right lateral shift. Nevertheless, panoramic and cone beam computed tomography (Fig 4) revealed a complete absence of the right condyle head and neck up to the level of the sigmoid notch. Panoramic radiographs from past records showed that at age 16 the superior aspects of the head of the left and right condyles were flattened but there were no signs or symptoms. The involvement of the TMJ in Satoyoshi syndrome is consistent with another case report whereby a young male had repeated TMJ dislocation.⁸

While the signs of condyle degeneration may be related to the syndrome, contributing factors in the present patient may include overloading of the TMJ as a result of loss of posterior occlusal support (subsequent to the tooth loss) along with strong contractions of the masseter muscles.¹⁹ Therefore, timely medical care is crucial for reducing muscle spasm and TMJ inflammation, and timely dental treatment with dentures is crucial for the restoration of posterior occlusal support to minimize overloading and progressive damage of the TMJ, muscles, and surrounding tissues.

Conclusions

Satoyoshi syndrome is a rare, progressive, debilitating condition of unknown etiology. In the orofacial region, it can manifest as progressive alveolar bone loss and root resorption that, along with excessive or repeated loading of teeth and joints subsequent to involuntary muscle spasms, can lead to premature tooth loss and degenerative changes in the TMJ. Considering the unknown etiology of the syndrome and the progressive nature of associated signs and symptoms, early diagnosis and long-term multidisciplinary treatment are crucial for preventing or minimizing complications and/or stabilizing already existing conditions by controlling contributing factors such as periodontal health, endodontic status, and occlusion.

Acknowledgments

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