Orthodontic treatment of a patient with Duchenne muscular dystrophy and macroglossia: How informed consent was critical to success

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This article describes the complex orthodontic treatment of a 22-year-old patient with Duchenne muscular dystrophy and macroglossia. His orthodontic treatment hinged on providing proper informed consent and management of the malocclusion with glossectomy, extractions, fixed appliances, and elastics. Challenges to traditional treatment are outlined, and compromises to both process and outcome are discussed from an informed consent point of view because of the serious risks involved. The treatment objectives were met, and the outcome was considered a success. (Am J Orthod Dentofacial Orthop 2013;144:890-8)

The purpose of this article is to describe the orthodontic treatment of a 22-year-old man with Duchenne muscular dystrophy and macroglossia. He used a power wheelchair that he controlled with a joystick, and some aspects of diagnosis and treatment were adapted to address his needs and abilities. I report here the treatment we provided, including the compromises that were made and the problems that arose. I discuss the patient's treatment based on his wishes and desires from an informed consent perspective and outline our limitations to "standard" orthodontic care delivery because of the unique nature of Duchenne muscular dystrophy.

DIAGNOSIS AND ETIOLOGY

A 22-year-old man came to the University of Minnesota faculty practice with a chief complaint of difficulty chewing; he reported that he was unable to bite into or chew his food effectively. He conveyed great frustration with this quality-of-life limitation to functional chewing. He was referred by his pediatric dentist and had no evidence of tooth decay or periodontal disease. He was accompanied by his mother and a caregiver. His past medical history was remarkable for Duchenne muscular dystrophy and an allergy to Augmentin. He did not have a tracheostomy tube. He was unable to voluntarily lift his arms and relied on caregivers for oral hygiene. The clinical examination and initial photographic montage (Fig 1) in full occlusion showed generalized excessive buccal crown torque with an anterior open bite of 8 to 10 mm and a posterior open bite of 0 to 12 mm. There was generalized mandibular spacing and an estimated 50% Class II molar relationship. He displayed signs of massive macroglossia and a single-point contact in maximum intercuspal position. He had only 50% incisor display on smile, and there was not a detectable centric relation to centric occlusion shift or discrepancy.

A medical computed tomograhpy scan was obtained from Suburban Imaging in Minneapolis. DICOM data were extracted and read at the University of Minnesota with help from an oral radiologist using volumetric, panoramic, and cephalometric reconstructions (Fig 2). These added to our problem list. We discovered an impacted maxillary right third molar and a supernumerary maxillary left impacted paramolar. His maxillary incisors were proclined with a U1-SN measurement of 120.3°.

Skeletally, he had discrepant maxillomandbular measurements with an ANB angle of −2.2° and a Wits value of 7.7 mm. With his degree of generalized proclination throughout both arches and related mandibular opening rotation, these anteroposterior skeletal measurements were given minimal diagnostic weight; the overall assessment from all records, including the clinical examination, suggested that his skeletal anteroposterior position was Class I.

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Fig 1. Pretreatment photographs.

The fact that he was occluding only on the lingual cusps of the left second molars (single-point contact) in addition to the generalized excessive buccal crown torque and mandibular generalized spacing led to the conclusion that these findings were sequellae from the macroglossia.

TREATMENT OBJECTIVES

The treatment goals in prioritized order were to (1) extract all third molars and the supernumerary paramolar, (2) reduce his massive tongue volume, (3) establish a functional occlusion, (4) close his anterior and posterior open bites and generalized spacing, and (5) increase his incisor display on smile.

TREATMENT ALTERNATIVES

The following treatment options were discussed with the patient.

1. No treatment.
2. Glossectomy with extraction of the supernumerary maxillary left molar and all third molars, wait 6 months for natural uprighting of the dentition, and reassess the patient for either premolar extractions or a nonextraction treatment plan; oral hygiene instructions for the caregivers; bond fixed appliances; level and align the arches using elastics to manage the severe proclination; and detail and finish occlusion until the objectives were met.

Although numerous glossectomy techniques have been described, the keyhole technique with anterior wedge reduction is the most common.[1] To establish a functional occlusion and close his bite, we intended to add simple crown tipping and lingual crown torque in a generalized fashion in both arches. With incisor uprighting, both bite closure and increased incisor display are known to occur through relative extrusion.[2] Some bite closure can be expected through molar uprighting as well; as the palatal cusps are tipped lingually, elimination of cuspal interferences should lead to bite closure through autorotation of the mandible. The mandibular interproximal spaces were to be closed with elastomeric chains.

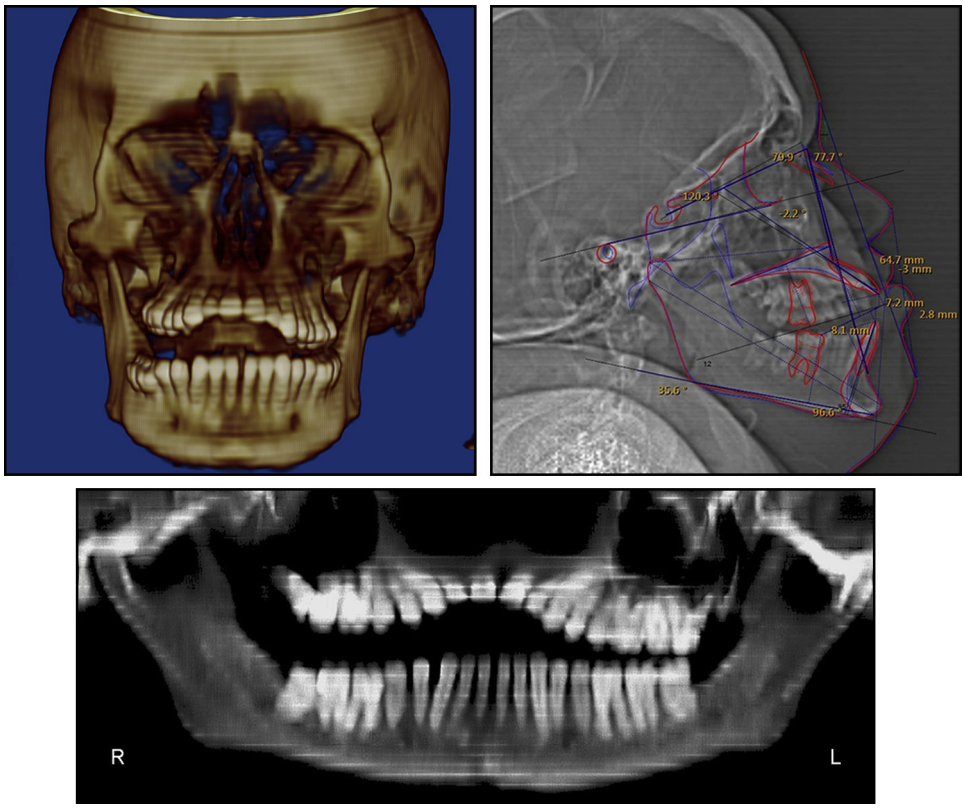

Fig 2. Medical computed tomography scan was used to generate cephalometric and panoramic reconstructions.

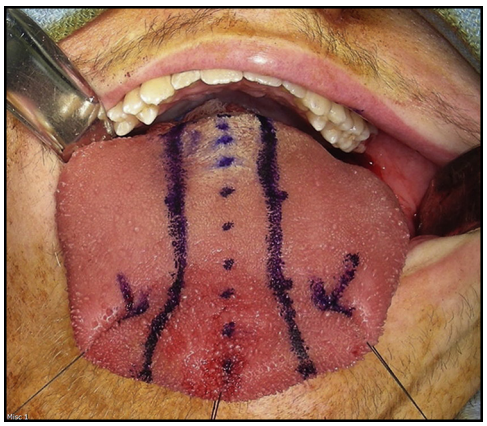

Fig 3. Keyhole wedge reduction glossectomy.

The patient and his parents were present during the informed consent and consultation appointment in March 2009. His chief concern was confirmed, his orthodontic findings were fully disclosed, his medical history was reviewed, and we discussed the above options for correction, including no treatment. We explained that although orthodontic treatment would not be stable unless the glossectomy procedure was performed, he was under no obligation to have treatment. Because of his strong commitment to improve his occlusion, he was sent for an evaluation for this glossectomy procedure and given a recommendation that he ask his other doctors about the inherent risks of the surgery in light of his preexisting medical condition. He was informed that his surgeon and medical doctor would be in the best positions to comment on these risks. The patient, under his own free will and with support from his parents, decided to go forward with the second treatment alternative above.

TREATMENT PROGRESS

In June 2009, the third molars were extracted, including the impacted supernumerary molar, and a keyhole wedge reduction glossectomy (Fig 3) was performed with the patient under general anesthesia. During the surgery, a tracheostomy procedure was also performed and a tracheostomy tube was placed. He spent approximately 1 month in a care facility after the surgery.

The patient returned to the University of Minnesota faculty practice for photos in January 2010, 7 months

Fig 4. Progress photographs, January 2010, 7 months after the glossectomy procedure and 2.5 months prior to fixed appliance placement. The 8-mm anterior open bite had closed spontaneously by approximately 6 to 7 mm, and the 12-mm posterior open bite on his right side closed by approximately 3 to 6 mm.

postsurgery, with a tracheostomy tube in place. The clinical examination showed that his 8-mm anterior open bite closed spontaneously by approximately 6 to 7 mm, and his 12-mm posterior open bite on the right side closed by approximately 3 to 6 mm (Fig 4). His hygiene was poor. The patient indicated that he still wanted braces for better chewing function, even though his chewing ability was improved from the surgery. A nonextraction plan was chosen for 2 reasons: (1) the dramatic bite closure observed in the 7 months immediately after his surgery and (2) the clinical estimation that our treatment goals could be achieved without extractions.

In April 2010 he demonstrated dramatically improved hygiene. Victory series (3M Unitek, Monrovia, California) 0.022-in slot Miniature Twin brackets were placed from first molar to first molar in both arches in addition to lingual attachments on the maxillary first molars and second premolars. Nickel-titanium archwires (0.014 in) were placed, and the patient was instructed to wear vertical Class III elastics to maintain positive overjet and "over the arch" cross elastics to upright and tip the mandibular molars (Fig 5). In June 2010, 0.017 × 0.025-in nickel-titanium archwires and transpalatal elastics along with continued cross elastics were placed for improved torque. In July 2010, his elastics were altered to mandibular transarch and vertical elastics. In August 2010, 0.018-in nickel-titanium archwires were completely engaged into the Miniature Twin slots to derotate his mandibular premolars. Dramatically improved bite closure and excellent uprighting were observed, so the elastic scheme was altered again for continued bite closure and molar buccal crown tipping. In October 2010, a 0.021 × 0.025-in nickel-titanium archwire was placed for finishing torque expression in his maxillary arch. A 0.016-in stainless steel archwire was placed to level his mandibular curve of Spee. In December 2010, 0.017 × 0.025-in maxillary and 0.016 × 0.022-in mandibular stainless steel wires were placed to finish leveling and continue torque expression. The use of vertical elastics dramatically improved his occlusion and provided a positive overbite.

In January 2011, treatment progress and goals were reassessed. Finishing goals were discussed with the patient and his parents. He was informed that the objectives of (1) improved biting and chewing function, (2) closing his anterior and posterior open bites, (3) closing his mandibular spaces, and (4) improving his anterior esthetics had been met. The patient inquired whether his bite could be improved; it could. He was told that we would need to upright his second molars, and it might take up to 1 more year; we reiterated the risks and benefits involved. After our full disclosure of his condition, the patient's wishes were to finish treatment to the most ideal occlusion possible. Brackets were then placed on his mandibular and maxillary second molars. A posterior open bite began to develop. It was determined to be cuspal and transitory and was managed with posterior transpalatal elastics across the maxillary molars and with vertical posterior configurations. In June 2011, the posterior open bite showed signs of improvement, so

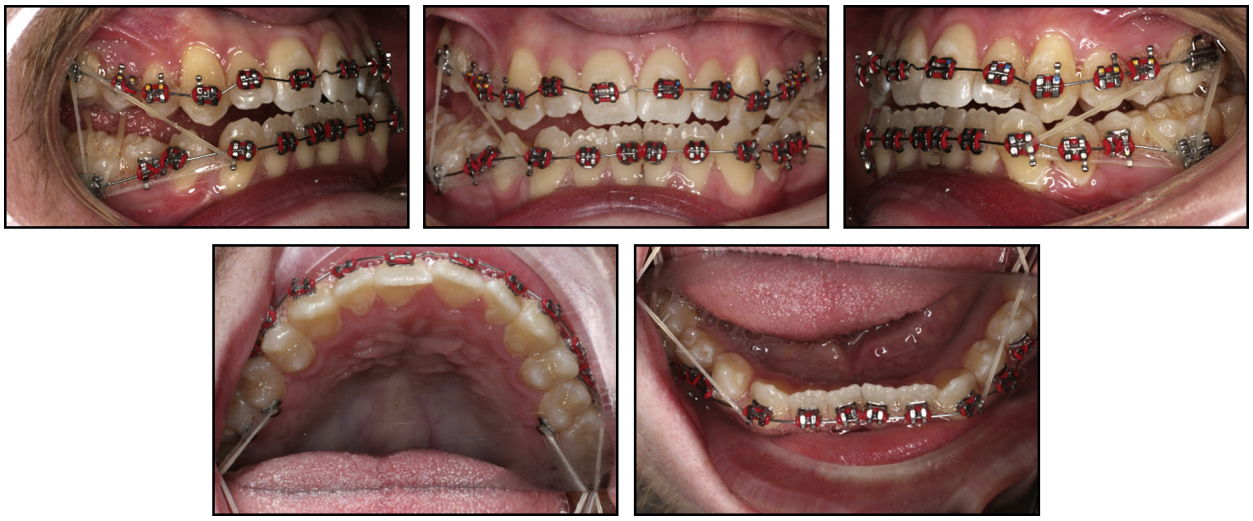

Fig 5. Progress photographs after placement of the 0.014-in nickel-titanium archwires. Vertical Class III elastics were prescribed to maintain positive overjet and over-the-arch cross elastics to upright and tip the mandibular molars.

transpalatal elastics were worn from mandibular molar to molar in addition to posterior box elastics for lingual crown torque expression and further bite closure (Fig 6). In July 2011, a posterior transverse discrepancy was observed on his right side that appeared clinically as though it could not be corrected without a greater time investment, more invasive mechanics, or surgery. Therefore, the decision was made to section the maxillary archwire between the maxillary right first and second premolars and leave his right side in a posterior lingual crossbite. One final adjustment was performed in September 2011. His maxillary second molars were in an improved and functional position. With his consent, the braces were removed, and Hawley retainers were placed in both arches. He was instructed to wear them full time. Final photos were taken in December 2011. The total time from the glossectomy procedure to fixed appliance placement was 9.5 months. His total time in active fixed appliances was 21 months (Fig 7). At 1 year postretention, there was minimal posterior open bite relapse (<1 mm) on his right side. His initial Hawley retainers still fit well, and he continued to wear them every night.

TREATMENT RESULTS

A functional occlusion was observed, and the patient reported much-improved chewing and biting. His anterior open bite was completely closed and remained closed for 13 months after removal of his appliances. His posterior open bite opened slightly on his right side, although he has not reported problems with mastication or esthetics. His mandibular spacing remains completely resolved. His incisor display increased from 50% on smile at the beginning of treatment to 90% after treatment. He was left with a unilateral posterior crossbite on his right side. He is pleased with the function and esthetics of his teeth. The treatment was considered a success in that it addressed his chief concern, and the treatment goals were achieved or surpassed.

DISCUSSION

Muscular dystrophy is a group of inherited diseases that cause progressive weakening of the voluntary muscles. Of the muscular dystrophies, Duchenne is the most common type seen in children. Duchenne affects only males and affects 1 in 3300 boys.[3] Onset is characterized by progressive muscle weakness in early childhood; patients usually need a wheelchair early in their second decade of life. With loss of function, the spine, arms, and legs become progressively deformed, and some cognitive impairment might be evident. Late stages of the disease are marked by labored breathing, cardiac problems, and ultimately death. Life expectancy has been increasing in recent years. From 1977 to 1984, the mean age of death was 18.9 $\pm$ 4.1 years; from 2003 to 2010, the mean age of death was 31.1 $\pm$ 5 years, and it appears to be rising.[4]

We encountered serious limitations to traditional record collection, but a concerted effort was undertaken to obtain a minimal starting diagnostic database nonetheless. Traditional imaging was not available for our patient because of the wheelchair design and its

Fig 6. Progress photographs, June 2011. The posterior open bite showed signs of improvement, so transpalatal elastics were worn from mandibular molar to molar in addition to posterior box elastics for lingual crown torque expression and further bite closure.

interference with our panoramic and cephalometric moving parts. Nearby dental imaging facilities did not have a Hoyer lift and were unwilling or unable to take panoramic or traditional lateral cephalometric radiographs. We ultimately sent him to a medical imaging facility for a computed tomography scan. With the help of an oral and maxillofacial radiologist, we obtained volume, panoramic, and cephalometric digital reconstructions (Fig 2). The patient and his parents expressed concern with the dental impressions and the potential choking risk. We therefore elected to forgo study models and instead focus our efforts on analyzing his photographs, computed tomography reconstructions, and clinical parameters. At the time, digital impressions were not in widespread use. We did not have a digital intraoral scanner, but this would be a good option for obtaining impressions if this treatment were started today. Bracket bonding and simple archwire adjustments were cumbersome. The wheelchair tipped back only about 45° from vertical, so we performed all intraoral procedures while standing, often on tiptoes or hunched over the patient in contorted and awkward positions to achieve basic access.

Equilibrium effects from the tongue, cheeks, and lips are well known to affect final tooth positions. Light but sustained soft-tissue pressures from the tongue are known to cause positional changes of the teeth.[5,6] Harvold[7] showed that reducing tongue volume on primates with hypertrophied tongues caused the arches to collapse lingually. Based on this body of work, we reasoned that the macroglossia was causing a pathologic disruption in dental equilibrium, resulting in his current presentation.

The purpose of informed consent is to protect our patients by ensuring that they are making informed decisions about the care they are about to receive. Informed consent requires 3 things: (1) full disclosure of the patient's condition by the doctor, (2) capacity of the patient to understand and appreciate the benefits and burdens of treatment, and (3) voluntariness, in that treatment commences only under the patient's own free will. Although doctors cannot legally and should not ethically lead their willing and trusting patients into harm's way, they are considered to have informed consent when these 3 conditions are met.[8]

Orthodontics is an elective specialty. It has been stated that the more elective the procedure, the greater the doctor's burden to inform the patient because the patient need not incur the risks.[9] Many studies have shown that patients want a high level of information before engaging in both nonelective[10-16] and elective[10,17,18] health care procedures. Recent research has clearly shown that adult orthodontic patients desire greater involvement with decision making as the orthodontic condition becomes more complex.[10] Informed patients tend to be better consumers of dental services. They have been shown to be less anxious, require fewer pain medications, have better postoperative pain control, and have more realistic expectations of their health care providers. They show better compliance and express greater satisfaction with the care that has been rendered.[19] For difficult cases like this one, these attributes of informed patients are more important than ever to a successful outcome.

Fig 7. Final photos, December 2011. Total time in fixed appliances was 21 months.

Informed consent is an integral aspect of orthodontic care; this case required a firm understanding of informed consent because the stakes to the patient's health and life were so great. He came to see us because he wanted to be able to chew his food better and to improve his quality of life. Based on our findings and previous research, a meaningful orthodontic result would not be possible or stable without a glossectomy.[20] However, surgery and the accompanying anesthesia pose real risks for a medically compromised Duchenne patient. As his orthodontist, I could not be in the position to accurately outline the risks involved with his surgical procedure. His surgeon, anesthesiologist, and respiratory therapist must carry this duty. Orthodontists are in the business of helping people with mostly low-risk, long-term, quality-of-life improvements. With high-risk patients such as this, it is incumbent upon us to understand in great detail what the patient truly wants as an outcome and why. Although I wanted to help him with his problem, I did not want or need the vicarious liability associated with this procedure; so, a referral was given to evaluate for a glossectomy procedure.[21] After the referral to his oral surgeon, it became apparent that the patient considered his potential for quality-of-life improvement worth these risks. During the surgery, a tracheostomy procedure was performed. The patient spent approximately 1 month in a care facility after the surgery. When we interviewed the patient, he stated that he felt that the surgery was worth it, despite the need for a tracheostomy procedure. His mother expressed gratitude that his tracheostomy procedure and placement was managed during the surgery and instead of at home, when he might have had a less ideal outcome or even death.

In hindsight, I would have managed two aspects of his orthodontic care differently. First, I would have bonded his second molars at the initial construction appointment. Instead of 21 months of treatment, the treatment might have finished earlier. My reasons for not bonding them initially were that (1) bracket placement was difficult so far back in the arch, (2) it was not critical to achieve our original treatment objectives, and (3) it was originally estimated that

treatment might be prolonged if they were incorporated early. Second, I would have attempted to correct his right-side transverse discrepancy earlier in treatment.

A statement from his mother in December 2012 indicated that he came down with severe bronchitis in June 2012 and was on a "cough-assist machine" to assist in expectorating excess mucus from his lungs. He reportedly has been doing much better with clearing his lungs, and the pneumonia has since resolved. Recently, his vallecula epiglottica "doesn't function very well," and he must be careful when eating; "he takes 2 small bites, drinks 2 sips, 2 small bites, etc." He reportedly still wears his Hawley retainers at night. He demonstrated some slight right-side posterior relapse but can still chew well and is pleased with his teeth. We consider this a successful outcome. The patient was due to graduate from technical college in June 2013.

CONCLUSIONS

The treatment of an adult with Duchenne muscular dystrophy has been described. The patient had limited physical abilities and used a heavy motorized wheelchair, which he controlled with a joystick. The records collection process and the basic orthodontic treatment delivery carried many logistic challenges and required a glossectomy procedure with serious medical risks. A firm understanding and appreciation of the principles of informed consent combined with a targeted realistic plan based on his chief concern guided our treatment to surpass fulfillment of the treatment goals and to help ensure a successful treatment and a satisfied patient.

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