Neuromuscular Disorders xxx (xxxx) xxx

Contents lists available at ScienceDirect

Neuromuscular Disorders

journal homepage: www.elsevier.com/locate/nmd

Picture of the month

Orthognathic surgery in RYR1-related congenital myopathy: a patient report

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ARTICLE INFO

Article history:
Received 24 May 2023
Revised 22 September 2023
Accepted 12 October 2023
Available online xxx

Keywords:
Orthognathic operation
Jaw surgery
Congenital myopathy
RYR1
CFTD

Patients with congenital myopathies often have characteristic dentofacial malocclusions that contribute to functional problems with feeding and drooling and psychosocial challenges. The Consensus Statement on Standard of Care for Congenital Myopathies by Wang et al. includes that surgical treatment of severe malocclusion should not be considered given the high risk of perioperative complications [1]. In contrast, a report on three patients with congenital myopathy suggested that orthognathic surgery can be carefully considered. These procedures typically require multidisciplinary pre- and postoperative evaluation and care over lengthy hospital stays with a high risk of respiratory complications that bear consideration in treatment planning [2].

The picture show the effects of the orthognathic operation in a now 40-year old patient with autosomal recessive RYR1-related congenital myopathy. She presented at birth with generalized muscle weakness, hypotonia, and ophthalmoplegia. Motor milestones were delayed. A muscle biopsy (at age 3) showed congenital fiber type disproportion. Genetic testing in adulthood (whole exome sequencing with panel analysis - DG-2.15, followed by RYR1 Sanger sequencing) had shown compound heterozygous RYR1 variants (c.10616G>*A* (p.(Arg3539His)); c.13033_13067del (p.(Ala4345fs)). Testing of asymptomatic parents showed that they were both carrier (mother of the VUS; father of the frameshift variant). These results, in combination with the results of the muscle biopsy and the phenotype had led to the diagnosis of autosomal recessive RYR1-related congenital myopathy.

Subsequently, her case was discussed in the multidisciplinary genetics – neurology – anesthesiology meeting. General anesthesia for a tonsillectomy in early childhood had been uneventful. The elective orthognathic operations had been performed without succinylcholine. Further details on the used anesthetic drugs were not available. Based on the normal results of invitro contracture testing of two unrelated individuals with the VUS, she was not considered to be at risk for malignant hyperthermia.

An orthognathic operation was considered in her teens, but postponed until 2012, in her late twenties. The indication for operation then was reduced orofacial function resulting in malnutrition (172 cm, 40 kg) and wish for cosmetic improvement.

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https://doi.org/10.1016/j.nmd.2023.10.005

Please cite this article as: A.J. van der Kooi, J. de Lange, M. Schouten et al., Orthognathic surgery in RYR1-related congenital myopathy: A patient report, Neuromuscular Disorders, https://doi.org/10.1016/j.nmd.2023.10.005

Fig. 1. The effects of the orthognathic operation in a now 40-year old patient with autosomal recessive RYR1-related congenital myopathy.
The indication for operation then was reduced orofacial function and wish for cosmetic improvement. The maxilla was moved forward and upward and the bone that was harvested during the upward movement was used to stabilize the widening of the zygomatic bones. Thereafter, the mandibula was shortened in order to achieve a good occlusion of teeth and a genioplasty was performed to improve the lip position and overall esthetic outcome.
A: Conventional X-ray lateral and anterior-posterior postoperatively.
B: Lateral photographs pre- and a few months postoperatively.
C: Series of portraits: pre-operatively; one year post-operatively; five years post-operatively and ten years post-operatively.

The process started with orthodontic treatment, alignment of the dental arches (age of 28).

The operation (age 29) lasted five hours. General anesthesia (without succinylcholine) was uneventful. Post-operative recovery was prolonged with fatigue, increased weakness and additional weight loss (3 kg). After she started using oral food supplements, she gradually improved in body weight (up to 55 kg) and general function. At age 40, she is very satisfied with the functional and cosmetic improvement (Fig. 1). This has increased her self-confidence enormously. She would like to encourage other patients to prepare the post-operative recovery well with a speech therapist and dietician.

Declaration of Competing Interest

None.

Acknowledgement

The authors are members of the Netherlands Neuromuscular Center and the European Reference Network for rare neuromuscular diseases (EURO-NMD). We are grateful to the patient who shared her experiences and composed this very illustrative figure. We acknowledge the RYR-1 Foundation for their continuous support for the worldwide patient community and the scientific research on RYR1-related myopathies.

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