Case Report

Multiple Parotid Masses: A Rare Case of De Novo Monomorphic and Pleomorphic Adenomas in a Patient with Myotonic Dystrophy

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Abstract
Objective/background. This is a case presentation of a primary multifocal monomorphic and pleomorphic adenoma, with possible association with myotonic dystrophy. Methods/presentation. The patient was a 36-year old woman with myotonic dystrophy with multiple left parotid masses. Biopsy and final pathology demonstrated discrete de novo pleomorphic and monomorphic adenomas. Conclusions/impact. There is increasing investigation into association between myotonic dystrophy and neoplasia, including salivary gland tumors. Here we present a patient with multiple parotid neoplasms and discuss this unique case in regards to the risk of tumors in patients with muscular dystrophy.

Keywords parotid mass; monomorphic adenoma; pleomorphic adenoma; myotonic dystrophy

1. Introduction

Pleomorphic and monomorphic adenomas are rarely multifocal. Reports of the incidence of de novo multifocal pleomorphic adenomas are in the range of 0.14%–0.6% [1]. Moreover, there are no reports of multifocal monomorphic adenomas and no reports of both pleomorphic and monomorphic adenomas arising within the parotid gland.

Myotonic dystrophy is a progressive muscular disorder, often starting in childhood or in the 20s, and progresses throughout life [2]. It is most noticeable in the hands and facial muscles, with ptosis, reduced facial expression, dysarthria, and gait abnormalities among the first signs and symptoms [3]. The most common form of myotonic dystrophy is caused by mutations in the DMPK gene with trinucleotide (CTG) repeat expansion leading to a dysfunctional gene product. Notably this disease is subject to anticipation, with increased CTG repeats in subsequent generations, resulting in earlier onset and more severe symptoms [3].

Interestingly, there are some case reports that suggest a relationship between myotonic dystrophy and cancer risk and tumor formation, including pleomorphic adenomas [4,5]. This relationship is hypothesized to arise from the genetic mutations and genetic instability (with unstable nucleotide triplet repeat sequences) contributing to myotonic dystrophy that could also play a role in tumorigenesis [4,6,7].

We present a patient with a history of myotonic dystrophy who displayed three distinct masses within the left parotid gland, ranging from 1.4 cm to 3.5 cm, arising from the superficial, deep, and parapharyngeal portions of the parotid gland.

2. Case presentation

A 36-year-old female presented with a left parotid mass that had been slowly increasing in size and tenderness over the course of two months. The patient had a history of deep venous thrombosis and adult-onset myotonic dystrophy, with facial, hand, and gait motor instabilities. She denied use of cigarettes or smokeless tobacco products and reported no alcohol consumption. Of note, she had a significant family history of myotonic dystrophy in her mother, maternal aunts, and grandmother, with earlier onset and worsening symptoms with each generation. Because of her strong family history, the patient had declined formal genetic testing. Notably, the patient had no history of prior fine needle aspiration, radiation, trauma or surgery of the head and neck.

On physical examination, a firm, 2–3 cm mass was palpated within the left parotid gland. An ultrasound-guided fine needle aspiration biopsy was performed with preliminary consistent with a pleomorphic adenoma, with no malignant cells identified. Computed tomography (CT) scans showed multiple discrete nodular enhancing lesions within the left parotid gland (Figures 1(a) and 1(b)). The deeper areas were not seen on ultrasound examination and were not biopsied. After discussion with the patient, these could be removed in a single surgery.

The patient underwent a left parotidectomy with facial nerve monitoring. Findings included a 1.4 cm circumscribed mass in the superficial lobe, a 3.5 cm circumscribed mass in
Figure 1: CT scans showing multiple discrete nodular enhancing lesions within the left parotid gland. (a) Parapharyngeal (arrow, 1) and deep lobe parotid tumor (arrow, 2). (b) A more inferior section of the deep lobe parotid tumor (arrow, 2) and the superficial parotid lobe tumor (arrow, 3).

the deep lobe of the left parotid gland, and a 3.5 cm circumscribed mass in the left parapharyngeal space. Each mass was completely excised with negative margins. The patient was seen back in follow up approximately one month after surgery. All incisions were well healed without complications. Final pathology results confirmed that the superficial mass was a pleomorphic adenoma and the two deep masses were discrete monomorphic adenomas (Figures 2(a)–2(c)).

3. Discussion

De novo multifocal salivary gland tumors are uncommon in general, and even more so in the case of pleomorphic adenomas and monomorphic adenomas [8]. There are no reports of the presence of both monomorphic and pleomorphic adenomas arising within the same gland. The majority of reported cases of primary multiple pleomorphic adenoma in previously untreated patients involve two dominant nodules; however, some have been reported with a larger number of nodules [9].

Given this patient’s history of myotonic dystrophy, the presented case is unique in the sense that it may provide insight into the potential relationship between germline mutations associated with myotonic dystrophy and the appearance of pleomorphic adenomas.

Myotonic dystrophy (a specific form of muscular dystrophy) is a multisystem disorder most commonly caused by a CTG expansion in the noncoding region of the DMPK gene, exhibiting effects on skeletal and smooth muscle, the eye, heart, endocrine system, and central nervous system [3,4]. This disorder can cause a number of symptoms ranging from muscle weakness to cardiac abnormalities [3]. Various neoplasms have been reported in patients with myotonic dystrophy. Most of these neoplasms involve the Wnt/β-catenin signaling pathway, suggesting

Figure 2: Histologic features of tumors isolated during a left parotidectomy; hematoxylin and eosin stained sections at 40× magnification. (a) Representative section of 1.4 cm pleomorphic adenoma isolated from the left superficial lobe. (b) Representative section of 3.5 cm monomorphic adenoma isolated from the left deep lobe. (c) Representative section of 3.5 cm monomorphic adenoma isolated from the left parapharyngeal space.
that tumor progression in myotonic dystrophy may relate to mutations in this pathway [4].

The association of pleomorphic adenoma and myotonic dystrophy has been previously described in only three case reports [10,11,12]. Johannesson and colleagues were the first to describe two patients with pleomorphic adenoma and myotonic dystrophy, and as they did not describe a mechanism, they argued statistically that the rarity of these two diseases makes their combination essentially impossible without an association [10]. Draper and Pickles expanded on this concept, reporting two further cases [11]. One case was a 9-year-old female with a single pleomorphic adenoma, and the second was a 29-year-old female with multifocal pleomorphic adenoma. Their conclusion was that these tumors are even more rare in children, suggesting an inherited relationship. Ogata confirmed this finding using southern blot analysis to show the parotid tumor, just as other abnormal functioning tissues in myotonic dystrophy (DM), had CTG repeats, suggesting a pathophysiologic mechanism of tumor formation [12]. Our case is unique in that there were three lesions noted in one patient, and that this is the first description of monomorphic adenoma as the pathology.

The probability of the repeated occurrence of these two conditions simply by chance is incredibly low; thus, physicians should be aware of the possibility of an association between myotonic dystrophy and salivary gland tumors, including pleomorphic and monomorphic adenomas. For patients with myotonic dystrophy, regular examination of the salivary glands may be indicated to identify potential masses. Further research into potential genetic drivers in pleomorphic and monomorphic adenomas arising in myotonic dystrophy patients, and comparisons of genetic profiles with those derived from nonmyotonic dystrophy patients may delineate both common and unique activated pathways in both pleomorphic and monomorphic adenomas.

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Conflict of interest The authors declare that they have no conflict of interest.

References