

# Diffuse Parotid Gland Enlargement Associated with Glycogen Storage Disease III: A Case Report

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The purpose of this study was to explore the patient whose diffuse parotid gland enlargement probable related with glycogen storage disease III (Cori-Forbes Disease). **Study Design and Setting:** A 13 year-old child who has a glycogen storage disease (GSD) III was referred to our department due to bilateral parotid gland swelling. We performed an open incisional biopsy of the parotid gland with continuous monitoring of the facial nerve. **Results:** Periodic Acid Shift (PAS) painting was studied to specimen and light microscopy of prepared was examined. The histological result was correlated well with parotid gland glycogenosis. **Conclusion:** Polysaccharide deposits in glycogenosis III are not restricted to skeletal muscle and liver, and to considering that polysaccharide accumulates to salivatory glands such as parotid gland and open incisional biopsy of the parotid gland can provide an accurate diagnosis, and can avoid the need for superficial parotidectomy for non-neoplastic or some disease associated with storage disorder This case is reported for salivatory, and especially parotid, gland glycogenosis have never been demonstrated before.

**Keywords:** Glikogen Storage Disease III, Diffuse Parotid Enlargement, Parotid Open Biopsy

## Introduction

Parotid gland enlargement duo to diffuse pathologic processes represents a diagnostic challenge for the surgeon. In such cases, fine-needle aspiration is inconclusive and superficial parotidectomy is usually performed. The smallest biopsy of the parotid gland generally performed is superficial parotidectomy (lateral lobectomy), the goal of which is to avoid tumor spillage and recurrence. However, in certain conditions, such as diffuse lymphoepithelial or lymphoproliferative disorders, an open biopsy of the parotid gland may be the diagnostic procedure of choice, as it involves minimal risk to the facial nerve(6).

In these cases, ultrasound, and MRI may also be inconclusive for definitive diagnosis. In this

situation, several authors argue that the role of open incisional biopsy for diagnosis and treatment.

Glycogenosis type III (Cori-Forbes disease) is an autosomal recessively inherited disease characterized by a deficiency in amylo-1,6-glucosidase or debrancher enzyme. Enzyme deficit can be detected in liver, heart, muscle, leukocytes, erythrocytes, and fibroblast and muscle cultures. Great amount of structurally abnormal glycogen are found principally in liver, muscle, and erythrocytes. Hepatomegaly is prominent in childhood. Muscle weakness can also be present (1).

Hepatic symptoms are decrease in adulthood. In adults, the clinical findings are dominated by muscle involvement, gradual weakness, exercise intolerance, and wasting of distal muscles (2-3). However, an adult patient with glycogen storage disease type IIIb who developed end-stage cirrhosis and a small hepatocellular carcinoma has reported by Elizabeth et al (7).

Cardiomyopathy has been reported in both children and adults (1-4). Recently, Schwann cell

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## CASE REPORT

involvement in peripheral nerve has been demonstrated by electron microscopy (1).

Sancho et al. had performed electron microscopy of skin specimens in four patients with glycogenosis III and revealed consistent abnormalities. They had observed massive glycogen storage in epithelial secretory cells of eccrine sweat glands and, less markedly, in smooth muscle fibers from the erector pili (1).

We thought in this case which abnormal glycogen might have been accumulated in parotid gland due to GSD III.

We reported a child 13 ages with bilateral diffuse parotid gland enlargement whom consistent abnormalities were found on light microscopy examination in parotid gland associated with glycogenosis.

### Case Report

We describe the case of a child 13 years old with bilateral diffuse parotid gland swollen who applies to our clinic last month. (Figure 1). The patient complained from preauricular swelling with painless and no fever and generally weakness, exercise intolerance and muscle involvement. There was these complaints accompanying with abdominal distention for ten years. She explained to be exposed repeatedly to the medical treatment with diagnosis of parotitis since five years old.



Figure 1. Bilateral Diffuse Parotid Gland Enlargement.

We doubt that glycogen storage disease and performed liver needle biopsy. It was concordant with abnormal glycogen accumulation in liver.

We observed the increase of SGOT(99), SGPT(109), LDH(324), GGT(77) enzyme levels on biochemical analysis. Sedimentation, erythrocytes, leukocytes and formulas were at normal levels (Figure 2). The evaluation of ultrasound was related with diffuse enlargement both of parotid and submandibular glands and there were any clear mass or nodule. At routine examine we did not clearly determine a mass in gland except for diffuse enlargement. We performed fine-needle aspiration (FNA) biopsy from gland but aspiration cytology outcome was inconclusive. It is widely accepted that a surgical biopsy should be performed if the FNA results are inconclusive. For this reason, we decided to take an open incisional biopsy of the parotid gland with continuous monitoring of the facial nerve.

ITEMS	LEVELS	RANGE
Leukocytes	5.5 K/uL	4.0-10.0
Erythrocytes	4.88 10 <sup>6</sup> /uL	3.60-5.70
Hemoglobine	14.1 g/dL	12.1-17.2
Hemotocytes	40.3 %	36.1-50.3
Sedimentation	11 mm/h	0-20
LDH	<b>324</b> u/L	125-243
SGOT	<b>99</b> u/L	5-34
SGPT	<b>109</b> u/L	0-55
GGT	<b>77</b> u/L	9-36

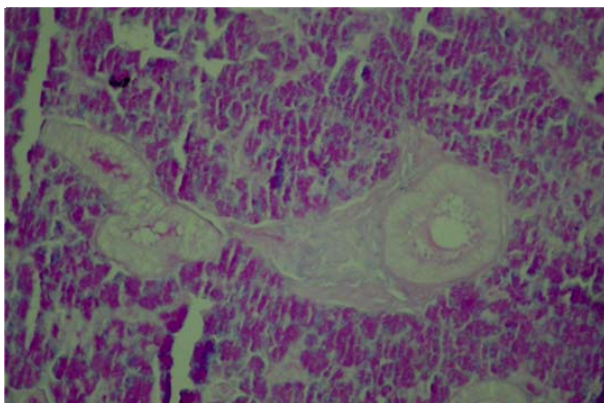
Figure 2. Biochemical analysis: Increases of liver function tests.

The parotid gland is closely applied to the posterior part of the ramus of the mandible, with the larger superficial lobe lying over the masseter muscle, and the deep lobe extending into the space poste-romedial to the ramus. Important anatomical structures pass through the gland, in particular the retromandibular vein and external carotid artery lie deep, while the facial nerve enters the postero-medial aspect of the gland and divides into branches which pass anterolaterally to emerge onto the anterolateral surface of the superficial lobe.

Passing diagonally across the gland, in the subcutaneous tissue, are branches of the great auricular nerve. The gland is enclosed by a dense well-defined capsule which is derived from the deep servical fascia (8).

We performed a surgical technique for open incisional biopsy from gland such as was describe by Ferguson et al technique (8). However, we used the facial nerve monitoring extra. Our technique is more reliable to avoid from facial nerve paralysis. 1 ml of a local lidocain solution is infiltrated about infraauricular region near the ear lobe. An incision, performed 1.5 cm in length, is placed in or paralel to a skin wrinkle immedately below or just behind and beneath the ear lobe, and deepened by blunt dissection to the gland capsule. A self-retaining retractor is placed and gland capsule is incised. We took a biopsy from gland about 5x5 mm.

Periodic Acid Shift (PAS) painting was studied to specimen and light microscopy of prepared was examined. The results of microscopic examination of specimens obtained in this study revealed a high proportion of exhibit parotid gland epithelial cells include dense glycogen deposits. Glycogen deposits was observed as lilac colour into gland epithelial cells with histochemically PAS painting x10 by a light microscopy (Figure 3).



**Figure 3.** Glycogen deposits are lilac colour in parotid gland epithelial cells with histochemically PAS painting x10 by a light microscopy.

## Discussion

Various pathological conditions may present with palpable swelling in the parotid region. These swellings may be neoplastic or benign, and intra-glandular or extra-glandular. Therefore histolo-

gical diagnosis weighs heavily on therapeutic decision making and prognosis (11).

Diffuse enlargement of the parotid gland is also present in other conditions such as sialolithiasis, bacterial or viral parotitis, Sjogren syndrome, or involvement by lymphoma, histological proof should be obtained for differantial diagnosis (12).

In this case we thought that polysaccharide might have been accumulated in parotid gland due to GSD III.

Ultrasound, FNA, and routine examiantion of parotid gland were not satisfactory for diagnosis.

As an alternative to superficial parotidectomy, open incisional biopsy of a parotid gland mass with facial nerve monitoring provides a good quality histological specimen without artifacts or obscu-ring blood, which are problems commonly associated with FNA (9-10).The routine use of facial nerve monitoring considerably increases the safety of such procedures, although they can be conducted under local anesthesia. The main objection regarding the performance of incisional biopsy of parotid tumor masses is the high probability of tumor spillage and recurrence. However, in diffuse pathologic conditions such as polysaccharide deposit in parotid gland likely our case, in which all or most of the gland is affected, tumor spillage is irrelevant.

Therefore, we treated open incisional biopsy to parotid gland with continuous monitoring of the facial nerve, too. Light microscopy of parotid specimens was performed with periodic acid shift (PAS) painting and massive glycogen storage was observed in gland. This study showed that glycolgen deposits in glycogenosis III are not restricted to liver, skeletal muscle and epithelial secretory cells of ecrine sweat glands of skin, in addition, polysaccharide deposits can also appearance in parotid gland.

We argued to need of open biopsy for diagnosis at some disorder like diffuse enlargement of parotid gland that this argument supported the studies of Sancho et al.

## Conclusion

In conclusion, this study was to show that glycolgen deposits in glycogenosis III are not restricted

to skeletal muscle and liver, and to considering that polysaccharide accumulates to salivatory glands such as parotid gland. Furthermore, it was argued the role of open incisional biopsy in some disorder at parotid gland likely diffuse enlargement and considered the polysaccharide storage in parotid gland with bilateral diffuse swelling.

Open incisional biopsy of the parotid gland can provide an accurate diagnosis, and can avoid the need for superficial parotidectomy for non-neoplastic or some disease associated with storage disorder.

Finally, it was recommended necessity more study about open incisional biopsy of parotid gland and especially diffuse accumulation of polysaccharide with swelling in parotid gland such as GSD III.

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