案例・5	Sex：♂	Age：8 month-old

Final anatomical diagnosis：

Glycogen storage disease, Type II (Pompe disease)

Hypertrophy, myocardium, severe, LV:17mm, RV:5mm, IVS:20mm in thickness, 200gm (normal 42.5gm).

Macroglossia

Hepatomegaly (340gm, normal 280 gm).

Vacuolar change of all skeletal muscle fibers.

Marked glycogen accumulations in: myocardia, liver, all skeletal muscle fibers and neurons of anterior horn of spinal cord.

Bronchopneumonia, lungs, bilateral, multifocal.

Effusions, peritoneal, 6 C.C.; pleural, right: 8 C.C.; pericardial, 17 C.C., all serosanguinous.

Hemorrhage, subdural, left temporal lobe; subarachnoid, left temporal and left frontoparietal.

Congestion, visceral, generalized

Disseminated intravascular coagulation (DIC), evidenced by small fibrinous microthrombi in lungs.

Abnormal lobulation, lung, right: 2 lobes, with absence of horizontal fissures.

Clinical summary：

This is a 8-month-old male infant died of hypertrophic cardiomyopathy and acute bronchiolitis. His birth history was unremarkable (G2P2, BBW : 2700GM, NSD) and had an elder brother who also died of hypertrophic cardiomyopathy at 5 month old.

He suffered from cough, poor appetite, restlessness and shortness of breath on Dec. 2000 when he was 5 month old and was brought to our OPD. Cardiac sonography was performed and hypertrophic cardiomyopathy with congestive heart failure (ejection fraction 54%) was found. He was admitted on Dec.12, 2000 , under the impression of cardiomyopathy with congestive heart failure and acute bronchiolitis. During admission, splenomegaly, protruded tongue and hypotonia were also noted on physical examination. The laboratory date showed : Lactate : 10.3, CK : 1262, MB : 2.8% , BB : 0% , MM : 97.2% ,AST : 377 and Pyruvate 0.29 . Therefore, Pompe's disease was suggested he was discharged on Dec. 26, 2000 , he was admitted again for acute bronchiolitis and stool impaction and was discharged o Jan. 6, 2001.

He suffered from fever, cough, rhinorrhea, poor activity and poor appetite since Mar. 19, 2001 and visited our emergency room. Physical examination showed coarse breath sound with rhonchi over bilateral lung fields. He was admitted to PICU under the impression of Pompe's disease, hypertrophic cardiomyopathy and R⁄O bronchopneumonia.

The chest X-ray showed cardiomegaly. Cardiac sonography revealed hypertrophic cardiomyopathy with the ejection fraction of 5%. The laboratory data showed GLU : 236 , AST : 393 , CRP < 2 , WBC : 12700 , SEG : 58% , HB : 11.2 . Oxygen, fluid restriction, digoxin, lasix and carnitine were used. The symptoms improved and he was transferred to ward on Mar. 20, 2001. However, cyanosis, bradycardia and no breath were noted on Mar. 22, 2001 and CPR was performed. He was transferred to PICU again. Due to poor response to CPCR, the patient expired on Mar. 22, 2001.

Autopsy findings :

(1) Body and external appearance :

Weight	6400 GM
Length	68 CM
Development	Fair
Nutritional state	Fair
Tongue	Macroglossia
Nail bed	Cyanosis
Circumference	Head : 41CM
	Thorax : 41CM
	Abdomen : 39CM

(2) Body cavities :

Pleural	Right	8 C.C. serosanguinous fluid
	Left	Unremarkable
Peritoneal		6 C.C. serosanguinous fluid
Pericardial		17 C.C. serosanguinous fluid

(3) Cardiovascular system :

Greater vascular structures	
Gross	Normal position

Heart General :			
Weight	200 GM	Pulmonary valve	2.48 CM
Left ventricle	1.7 CM	Mitral valve	3.7 CM
Right ventricle	0.5 CM	Right ventricle IVS	2.0 CM
Tricuspid valve	3.14 CM	Aortic valve	3.14 CM
Foramen ovale	Close	Aortic valve	Close

Cardiovascular system	Gross	Microscopic Diagnosis
Valves	Unremarkable	
Epicardium	Unremarkable	Unremarkable
Myocardium	Hypertrophic	Sections show cardiac muscle with cytoplasmic vacuolization
Endocardium	Unremarkable	Unremarkable
Coronary arteries	Unremarkable	

(4) Respiratory system :

Respiratory system	Gross	Microscopic Diagnosis
Trachea and major bronchi	Contents : No aspiration materials	
Mediastinum : Thymus	6 GM, 5.5 x 5.0 x 0.4 CM	Unremarkable

Lung						
Gross	Weight	Right	60 GM	Size	Right	10 x 9.0 x 1.5 CM
		Left	80 GM		Left	12 x 9.0 x 3.0 CM
	Congestion and consolidation					
Microscopic Diagnosis	Sections show congestion and focal accumination of neutrophils and eosinophils in the alveolar spaces. Multiple fibrinous microthrombi are seen in the small arterioles.					

(5) Gastrointestinal Tract :

Pharynx	Gross	Microscopic Diagnosis
	Unremarkable	

Esophagus	
Gross	Unremarkable
Microscopic Diagnosis	Sections show skeletal muscle with cytoplasmic vacuolization. There is acute and chronic inflammatory cell infiltration in the submucosal and muscular layers.

Gastrointestinal Tract	Gross	Microscopic Diagnosis
Stomach	Unremarkable	Unremarkable
Duodenum	Unremarkable	Unremarkable
Salivary gland	Unremarkable	Unremarkable
Small bowel	Unremarkable	Unremarkable
Large bowel	Unremarkable	Unremarkable
Cecum and appendix	Unremarkable	
Pancreas	Weight : 5 GM	Unremarkable
	Size : 7.0 x 1.0 x 1.0 CM	
	Unremarkable	

(6) Hepatobiliary system :

Liver		
Gross	Weight	340 GM
	Size	16.5 x 15 x 7.0 CM
	Outer surface	Smooth hepatomegaly
	Cut surface	Unremarkable
Microscopic Diagnosis	Section show hepatocytes with diffuse cytoplasmic vacuolization. PAS stain is strongly positive. Electron microscopic examination reveals glycogen deposition in multiple membrane-bound nodules. FAT stain is focally positive.	

Gallbladder, Bile ducts	Gross	Microscopic Diagnosis
	Unremarkable	Autolysis

(7) Hematopoietic-Lymphoid system :

Spleen		
Gross	Weight	20 GM
	Size	9.5 x 4.5 x 1.5 CM
	Unremarkable An accessory spleen measuring 1.2 CM and diameter is also found.	
Microscopic Diagnosis	Unremarkable	

Hematopoietic-Lymphoid system	Gross	Microscopic Diagnosis
Lymph nodes	Unremarkable	Unremarkable
Bone marrow	Unremarkable	Unremarkable

(8) Urogenital system :

Kidney						
Gross	Right	Weight	26 GM	Left	Weight	27 GM
		Size	5.5 x 4.0 x 2.5 CM		Size	5.5 x 2.5 x 2.5 CM
	Unremarkable					
Microscopic Diagnosis	Congestion					

Urogenital system	Gross	Microscopic Diagnosis
Ureters	Unremarkable	Unremarkable
Bladder and urethra	Unremarkable	Unremarkable

Prostate/Testes						
Gross	Right	Weight	1.0 GM	Left	Weight	1.0 GM
	Unremarkable					
Microscopic Diagnosis	Unremarkable					

Tongue	
Gross	Macroglossia
Microscopic Diagnosis	Sections show skeletal muscle with cytoplasmic vacuolization consistent with glycogen storage disease.

(9) Endocrine system :

Endocrine system	Gross	Microscopic Diagnosis
Thyroid	Weight : 2 GM	Unremarkable
	Unremarkable	
Pituitary	Unremarkable	Unremarkable
Adrenals	Right : 1 GM	Unremarkable
	Left : 1 GM	
	Unremarkable	
Psoas muscle	Unremarkable	Sections show skeletal muscle with cytoplasmic vacuolization consistent with glycogen storage disease.
Skin	Unremarkable	Unremarkable

(10) Central nervous system :

Central nervous system		Gross	Microscopic Diagnosis
Brain	Weight	783 GM	Marked ballooning and swelling of all the neuron cells in the spinal code, especially anterior horn. The posterior nerve roots also show similar change of neurons. The neuron cells and astrocytes in medulla are also involved. PAS stain are strongly positive in all these cells. The cerebrum shows no significant change.
	Shape size	13 x 11 x 10 CM	
Leptomeninges		Intact	
Cranial nerves		Intact	
Circle of Willis		Intact	
Spinal cord		Intact	
Cerebellum		95GM	

Central nervous system:	
Scalp	Unremarkable
Dura	Subdural hemorrhage, multifocal
Falx	Unremarkable
Tentorium	Unremarkable

Section Taken and Labeled :

Labeled	Section Taken	Labeled	Section Taken
1	Chest wall skeletal muscle	**27**	Lung, LUL
2	Liver	**28**	Lung, LLL
3	Heart	**29**	Liver, left lobe
4	Accessory spleen	**30**	Liver, caudate lobe
5	Carotid lymph node	**31**	Liver, right lobe
6	Psoas muscle	**32**	Gallbladder
7	Skin	**33**	Spleen
8	Pituitary gland	**34**	Diaphragm
9	Psoas muscle	**35**	Stomach, duodenum
10	Left ventricle	**36**	Left kidney and ureter
11	Right ventricle	**37**	Right kidney and ureter
12	Bone marrow	**38**	Urinary bladder
13	Pancreas	**39**	Mesentery lymph node
14	Pericardium	**40**	Salivary gland
15	Left adrenal	**41**	Thyroid
16	Right adrenal	**42**	Tongue
17	Left testis	**B1**	Right frontal lobe
18	Right testis	**B2**	Right parietal lobe
19	Thymus	**B3**	Right hippocampus
20	RA	**B4**	Right basal ganglia

Section Taken and Labeled :

Labeled	Section Taken	Labeled	Section Taken
21	RV	**B5**	Right temporal lobe
22	LA	**B6**	Right occipital lobe
23	LV	**B7**	Medulla
24	Aortic arch	**B8**	Midbrain
25	Lung, RUL	**B9**	Spinal cord
26	Lung, RLL	**B10**	Dentate nucleus
1, 2, 3, 9 and 10 fixed in alcohol		**B11**	Spinal cord

Final Comments :

This is a 8 month-old male infant will hypertrophic cardiomyopathy and died of cradio-respiratory failure. Autopsy findings mainly shows vacuolar change and marked glycogen deposition in all skeletal muscle, myocardium, liver and the neurons of spinal cord and medulla. The whole pictures are compatible with type II glycogen storage disease. Genetic study of the myocardium and skeletal muscle revealed mutation of the specific gene for acid-maltase at the exon 13. The lungs show mild bronchopneumonia multifocally. The latter should be secondary to severe heart failure and poor motor function .