# RESULTS

This study included 629 cardiomyopathic patients who were following up at the Pediatric Cardiology Clinic, ABO ELRESH Hospital, Cairo University during almost an eight years study period (from January 2003 to November 2010) with 94 of them were diagnosed as metabolic cardiomyopathy.

## DESCRIPTIVE DATA:

### 1) Demographic data of patients with metabolic cardiomyopathy:

***Table 9: Age of cases (in months).***

| | Mean | ±SD | Minimum | Maximum |
|---|---|---|---|---|
| **Age in months** | 3.1 | 4.8 | 1 month | 156 month |

SD: Standard deviation

This table shows that the mean age of the patients at diagnosis was 3.1 month.

***Table 10: Age groups.***

| | Number of patients (94) | Percentage 100% |
|---|---|---|
| **0 - 6 m** | 31 | 32.9% |
| **7m - 12m** | 9 | 9.5% |
| **1y - 5y** | 33 | 35.4% |
| **5y -10y** | 14 | 14.8% |
| **Above 10y** | 7 | 7.4% |

This table shows that the most common age group was 1-5 years (35.4%), then 0-6 month (32.9%) and the least common age group was above 10 years of age (7.4%).

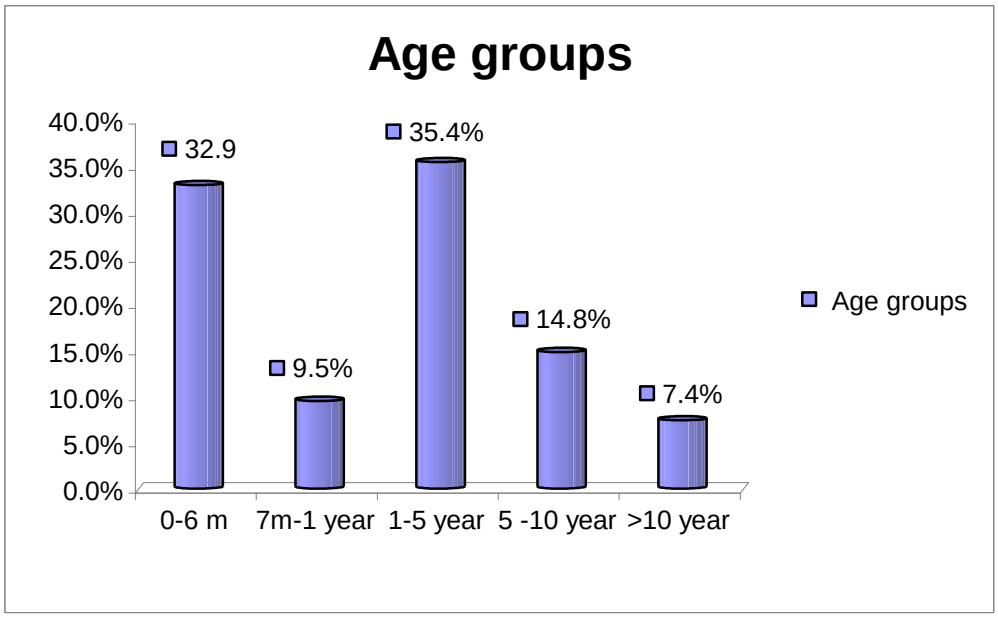


**Fig. (11): Distribution of Age groups in cases with metabolic cardiomyopathy**

***Table 11: Gender distribution of patients.***

| | | Cases | |
|---|---|---|---|
| | | N | % |
| Sex | Male | 59 | 62.7 |
| | Female | 35 | 37.3 |

This table shows that most of patients with metabolic cardiomyopathy were males with percentage of 62.7%.

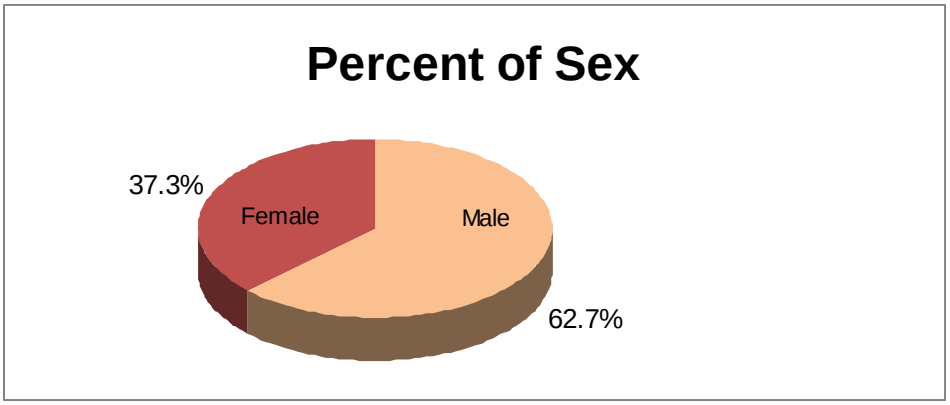


**Fig. (12): Distribution of sex among cases with metabolic cardiomyopathy.**

***Table 12: Family history.***

| | Cases | |
|---|---|---|
| **Family history** | **No (94)** | **100%** |
| **Consanguinity** | 25 | 26.6% |
| **Similar condition** | 8 | 8.5% |
| **Sudden death** | 2 | 2.1% |
| **Negative** | 59 | 62.8% |

In the present study, shows that, consanguinity was positive in (26.6%) of all cases. A history of affected similar condition was positive in (8.5%) while history of sudden death was positive in (2.1%) of metabolic cardiomyopathic patients.

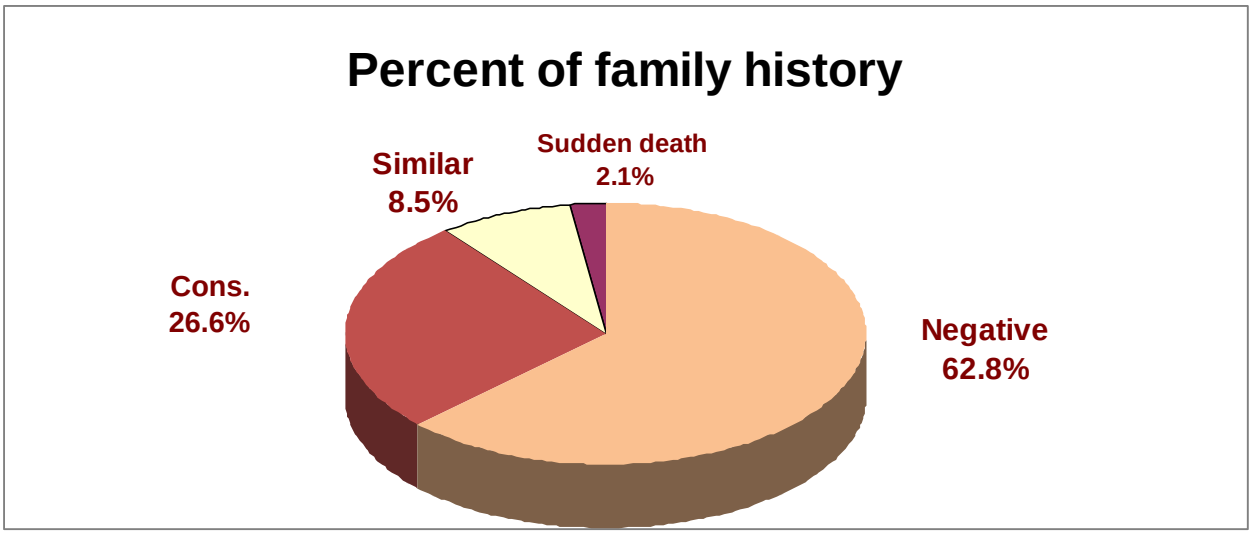


**Fig. (13): Distribution of family history in our study**

## *2) Clinical Presentation of patients with metabolic cardiomyopathy:*

***Table 13: Onset of illness.***

| | Cases | |
|---|---|---|
| ***Duration of illness*** | **No** | **%** |
| **Acute** | 53 | 56.3% |
| **Chronic** | 38 | 40.4% |
| **missed** | 3 | 3.3% |

This table shows that the majority of cases were presented acutely (56.3%) while other presentations were chronic illness (40.4%) with 3 missed cases not reported in the files.

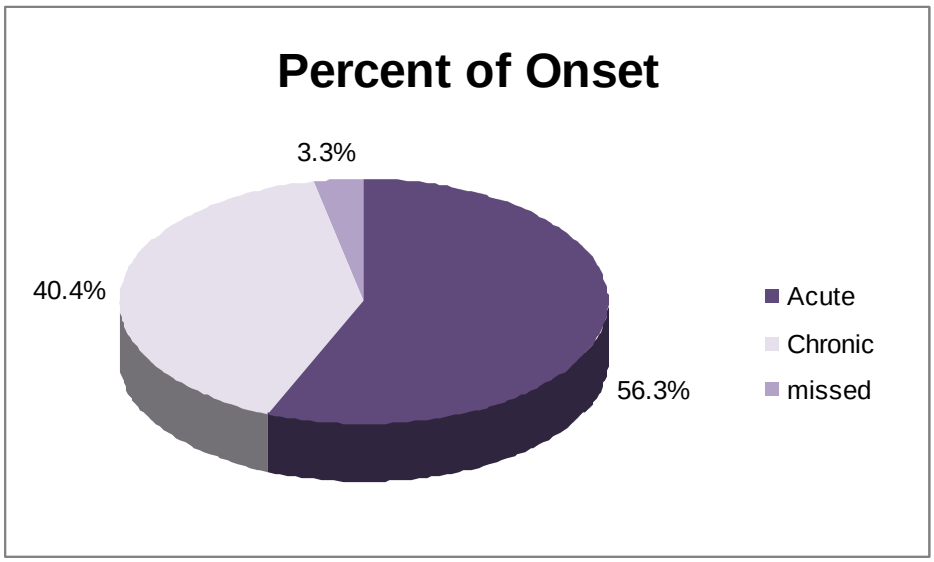


**Fig.(14): Distribution of onset in cases of metabolic cardiomyopathy.**

***Table 14: Clinical presentations.***

| | Cases | |
|---|---|---|
| ***Clinical presentations*** | **No** | **%** |
| **Cardiac symptoms (Heart failure, arrhythmia, recurrent chest infection)** | 73 | 77.6% |
| **Seizures** | 12 | 12.7% |
| **Gastroenteritis** | 9 | 9.7% |

This table shows that the majority of cases presented with cardiac symptoms (77.6%) while other presentation as seizures represents (12.7%) and recurrent GE represents (9.7%) of cases followed up at the clinic.

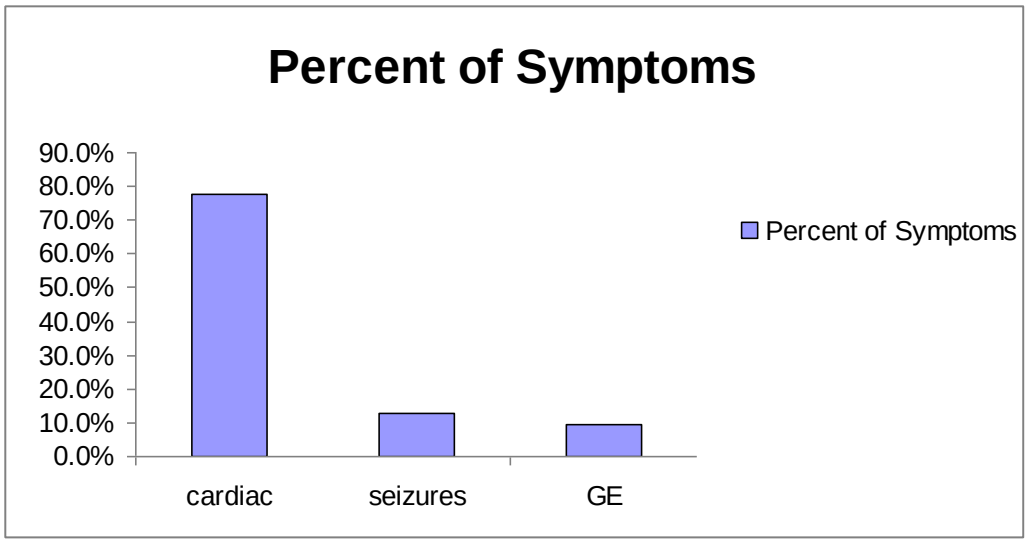


**Fig (15): Distribution of symptoms among cases with metabolic cardiomyopathy**

***Table 15: Extracardiac Manifestation.***

| *Extracardiac manifestation* | Cases | |
|---|---|---|
| | **No (63/94)** | **%** |
| **Seizures** | 28 | 29.7% |
| **Skeletal& coarse feature** | 8 | 8.5% |
| **Neurological (Hypotonia etc…)** | 7 | 7.4% |
| **Psychomotor regression** | 3 | 3.1% |
| **Mental Retardation** | 2 | 2.1% |
| **Hepatomegaly** | 2 | 2.1% |
| **Retinitis pigmentosa** | 1 | 1% |
| **Others** | 12 | 12.7% |

This table shows that 63 of cases had extracardiac manifestations, 28 of them had seizures (29.7%) &7 of them had neurological affection (7.4%).

## 3) Classification of cases with cardiomyopathy in our study:

***Table 16: Classification as regard Etiology.***

| Etiology of cardiomyopathy | Cases | |
|---|---|---|
| | No(629) | % |
| **Idiopathic** | 279 | 44.4 |
| **Myocarditis** | 146 | 23.2 |
| **Metabolic** | 94 | 14.9 |
| **Familial** | 74 | 11.7 |
| **Neuromuscular** | 50 | 8 |
| **Cardiac anomalies** | 26 | 4.1 |
| **Renal** | 23 | 3.7 |
| **Collagen** | 15 | 2.4 |
| **Endocrine** | 8 | 1.3 |
| **Hematology** | 2 | 0.3 |

This table shows that the most common cause of cardiomyopathy in our study is of idiopathic etiology (44.4%), cardiomyopathy due to myocarditis presented (23.2%), while metabolic etiology presented (14.9%) and familial (11.7%) of cases followed up at the clinic.

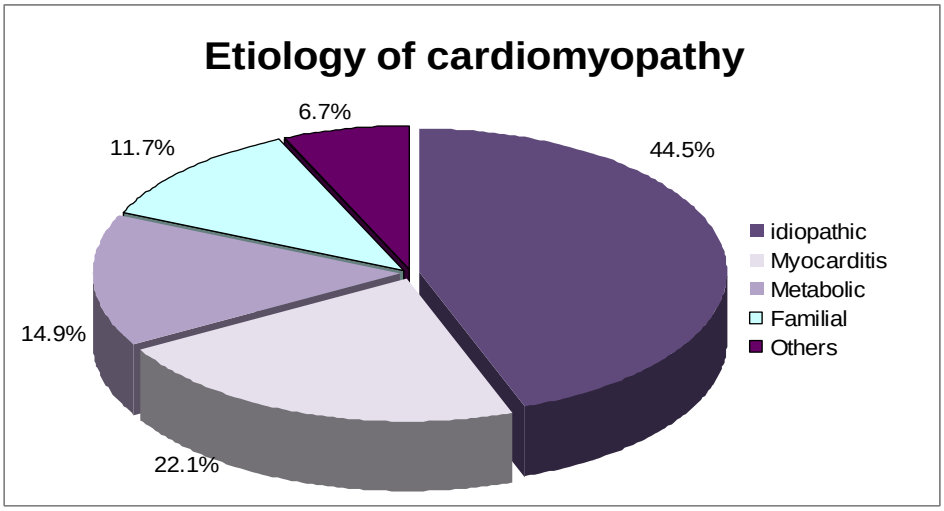


**Fig (16): Distribution of etiology of cardiomyopathy cases.**

***Table 17: Classification of the types of metabolic cardiomyopathy cases which were diagnosed by Echocardiography.***

| Types of Metabolic cardiomyopathy | No (94) | 100% |
|---|---|---|
| **Hypertrophic cardiomyopathy (HCM)** | 48/123 | 39% |
| **Restrictive cardiomyopathy (RCM)** | 4/28 | 14.2% |
| **Dilated cardiomyopathy (DCM)** | 39/457 | 8.5% |
| **Others (LVNC, Unclassified)** | 3/15 | 20% |

This table shows that the majority of cases of metabolic causes were HCM (39%) of total cases of HCM. DCM cases of metabolic causes were presented in (8.5%) of total cases of DCM and RCM cases were presented in (14.2%) of total cases of RCM followed up at the clinic.

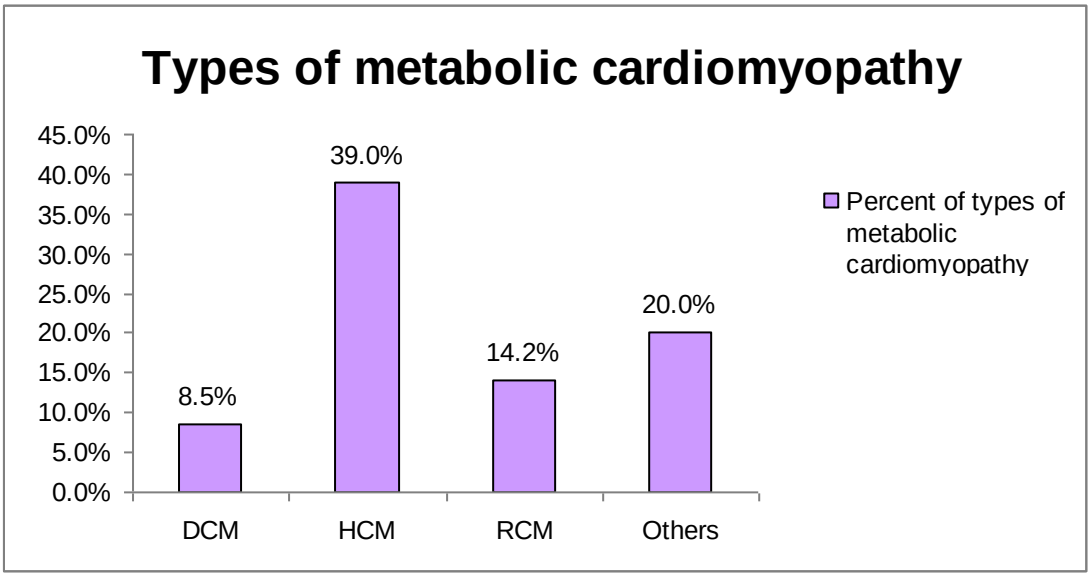


**Fig. (17): Types of metabolic cardiomyopathy cases.**

***Table 18: Classification of metabolic cases in our study as syndromes &Query cases.***

| | Cases | |
|---|---|---|
| | **No (94)** | **100%** |
| **Query Metabolic** | 63 | 67% |
| **Metabolic syndromes (GSD, MPS, L-carnitine deficiency, Mitochondria)** | 31 | 33% |

This table shows that 31 cases (33%) have been diagnosed as syndromes of metabolic cardiomyopathy while 63 cases (67%) were highly suggestive of metabolic etiology for cardiomyopathy.

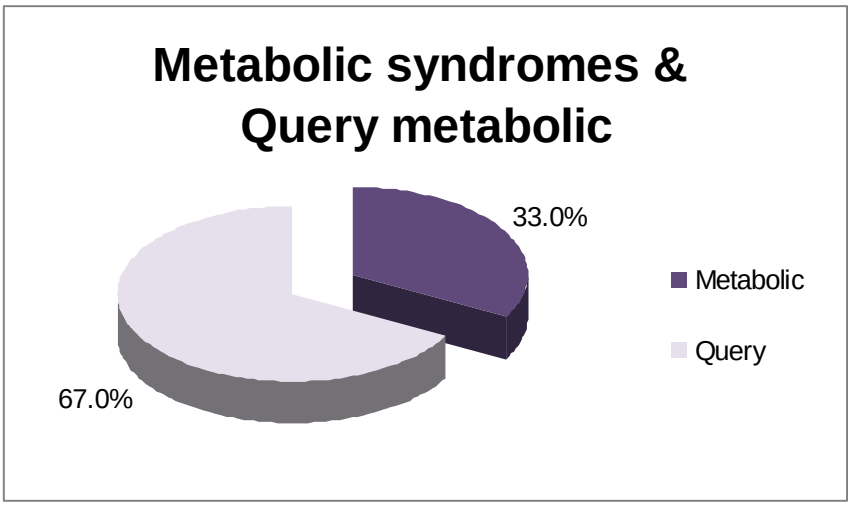


**Fig. (18): percent of metabolic syndromes with cardiomyopathy &Query metabolic in our study**

***Table 19: Classification of metabolic syndromes.***

| **Metabolic Syndromes** | **Cases** | |
|---|---|---|
| | **No(31/94)** | **33%** |
| **Lysosomal storage disease(MPS)** | 13 | 13.8% |
| **L-Carnitine deficiency** | 13 | 13.8% |
| **GSD** | 3 | 3.2% |
| **Mitochondria** | 2 | 2.2% |

This table shows that the majority of syndromes of metabolic cardiomyopathy were MPS (13.8%). L-Carnitine deficiency was presented in (13.8%) of cases. GSD was presented in (3.2%) of cases followed up at the clinic.

***Table 20: L-carnitine deficiency in cases with metabolic cardiomyopathy.***

| | Cases | |
|---|---|---|
| **L-carnitine deficiency** | **patients No (94)** | **percentage** |
| **Deficient** | 13 | 13.8% |
| **Normal** | 25 | 26.7% |
| **Missing** | 56 | 59.5% |

This table shows that serum L-carnitine was done for 38 patients of 94 patients who were diagnosed as metabolic cardiomyopathy, thirteen of them had L-carnitine deficiency represented (13.8%) with missing 56 cases were not reported in the files.

As regard other laboratory investigation that were done as a part of screening for metabolic cardiomyopathy, such as blood studies of acyl glycine derivatives and quantitative organic acids, ammonia, lactate, pyruvate, (23/94) patients had done these labs. It revealed elevated Glycosamineglycans (GAGs) in 2 out of 11 patients. Cetyle pyridinium chloride (CPC) was elevated in 2 out of 3 patients; lactate was elevated in 1 out 3 of patients.

## 5) X-ray finding:

Concerning the X-ray findings in our study, the available films were found in 92 cases. Cardiothoracic ratio was increased in 87 cases of them (94.5%), 13 cases were (14 %) showed pulmonary congestion, 9 cases were associated with chest infection (9%) and 1 case (1%) had pleural effusion..

## 6) ECG finding:

There were only 45 available ECGs, only (11%) of them had irregular sinus rhythm. Heart rate was abnormal as regard age in 33% of patients presenting with tachycardia, only (35.5%) revealed ventricular enlargement, (26.6%) revealed axis deviation while abnormal ST segments occurred in about (17.7%) of cases.

## 7) Echo finding:

Concerning the Echocardiographic findings in our study, the available echocardiograms (ECHOs) were found in 94 cases. As regard types of cardiomyopathy 48 cases of them (39%) presented with hypertrophic cardiomyopathy, 39 cases (8.5%) presented with dilated cardiomyopathy, 4 cases (14.2%) presented with restrictive cardiomyopathy and 3 cases (20%) presented with other types of cardiomyopathy (2 cases of them were left ventricular non compaction and 1 case was unclassified).

## *8) Medication:*

The medications received were mainly for treatment of heart failure with (57.2%) of patients were on diuretics, (36.4%) were on Digoxin (55.2%) were on ACEI while 18% of patients were on L-carnitine therapy (13.8% of them were L-carnitine deficiency syndrome and remaining 4.2% were query metabolic without improvement).