

## **Cystic Fibrosis In Jordan : Clinical And Genetic Aspects**

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**Objective:** To describe the demographic characteristics, phenotype, genotype, microbiological data, treatment and outcome among Jordanian children with cystic fibrosis .

**Design:** A prospective Cohort Study .

**Setting:** Princess Rahma Teaching Hosptial (PRTH).

**Methods:** All patients with cystic fibrosis seen between 1995 and 2000 inclusive (n=72). Clinical and laboratory data were collected on these patients.

**Results:** There were 37 males and 35 females in the study group. The mean age of all patients was 4.5 years; the mean age at presentation was 21.2 months; the mean age at diagnosis was 30.7 months and the mean delay of CF diagnosis was 9 months. Pancreatic exocrine insufficiency was documented in 94% of cases. Twenty (27%) children died, most below the age of 1 year. Pseudomonas aeruginosa was isolated in 30% of cases. Therapeutic measures were suboptimal in the majority of cases.

Consanguineous marriage was present in 70% of cases. Genetic screening of the study population revealed 20 different Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) mutations with Delta F508 mutation accounting only for 6.3%.

**Conclusion:** There is a wide variability in the phenotype and genotype of patients with cystic fibrosis. Jordanian CF patients have a severe clinical course of disease with genetic and environmental factors may be contributory.