History of CF in Turkey

Elif Dagli

Istanbul Turkey

1963- Clinicopathological conferences Hacettepe- Ankara

Cystic fibrosis was a post-mortem diagnosis.

1963 Ankara

First article

Helv Paediatr Acta. 1973 May;28(2):165-74.

Newborn screening for cystic fibrosis with the chloride electrode and neutron activation analysis.

Gürson CT, Sertel H, Gürkan M, Pala S.

High school

Development of Pediatric Pulmonology

1974-Hacettepe University Department of Pediatric Pulmonology

Brompton Hospital 1988-1990

Hum Genet. 1990 Sep;85(4):409-10.

Frequency of the F508 deletion in the CFTR gene in Turkish cystic fibrosis patients.

Hundrieser J[1], Bremer S, Peinemann F, Stuhrmann M, Hoffknecht N, Wulf B, Schmidtke J, Reiss J, Maass G, Tümmler B.

Abstract

The F508 deletion in the **cystic fibrosis** transmembrane conductance regulator (CFTR) gene was found in 8 out of 30 Turkish **cystic fibrosis** (CF) chromosomes (27%). Five Turkish delta F508 CF chromosomes were associated with the risk haplotype B in KM19 (2 allele)/XV2c (1 allele). In the Turkish population, **cystic fibrosis** is predominantly caused by mutations other than the F508 deletion.

Parental consanguinity

Development of Pediatric Pulmonology

1990 -Marmara University Department of Pediatric Pulmonology

1992 -Turkish Thoracic Society Pediatric Pulmonology Working Group

1992 –Egean University Department of Pediatric Pulmonology

2

Muhammed
At 1 year

“On 17th July 1989 it was very hot at the seaside. My 6-year-old son fainted, admitted to the intensive care. No body could explain why he collapsed, but recovered in 2 days. He was sweating a lot, salt granules were accumulating on his skin. My family physician recommended sweat test. It was not performed properly anywhere. They were putting the child’s hand into a plastic bag and waiting the the sweat”

“We decided to go to Netherlands for further investigations. The doctors there did not think of CF and refused to do the sweat test. I insisted and the test result was positive”

“We returned to Turkey and tried to find a doctor. Nobody knew anything about the disease. Pediatricians were telling me that it was not an important disease just causing random cysts”

“ I read an article about cystic fibrosis in a Turkish popular medical magazine called Dirim. The author was trying to increase awareness about the disease and asking the patients to apply to Marmara University. I found her.

My son will be 35 years old on 21st March. He is married fo 2.5 years.”

Turk J Pediatr. 1991 Apr-Jun;33(2):79-84.

Prenatal diagnosis of cystic fibrosis in a Turkish family.

Yilmaz E[1], Ozgüç M, Coşkun T, Beksaç S, Cakar N, Ayter S, Ozalp I.

Ann Genet. 1993;36(3):144-9.

Molecular genetic analysis of Turkish cystic fibrosis patients.

Köprübasi FF[1], Malik N, Bösch-al-Jadooa N, Alkan M, Tanac R, Bühler E.

Eur J Pediatr. 1994 Nov;153(11):829-31.

Sodium chloride deficiency in cystic fibrosis patients.

Ozçelik U[1], Göçmen A, Kiper N, Coşkun T, Yilmaz E, Ozgüç M.

Hum Hered. 1995 May-Jun;45(3):175-7.

Study of 12 mutations in Turkish cystic fibrosis patients.

Yilmaz E[1], Erdem H, Ozgüç M, Coşkun T, Ozçelik U, Göçmen A, Ozalp I.

Hum Genet. 1996 Jun;97(6):727-31.

Geographic distribution and origin of CFTR mutations in Germany.

Tümmler B[1], Storrs T, Dziadek V, Dörk T, Meitinger T, Golla A, Bertele-Harms RM, Harms HK, Schröder E, Claass A, Rutjes J, Schneppenheim R, Bauer I, Breuel K, Stuhrmann M, Schmidtke J, Lindner M, Eigel A, Horst J, Kaiser R, Lentze MJ, Schmidt K, von der Hardt H, Estivill X.

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The State of Cystic Fibrosis in Turkey

Authors Authors and affiliations

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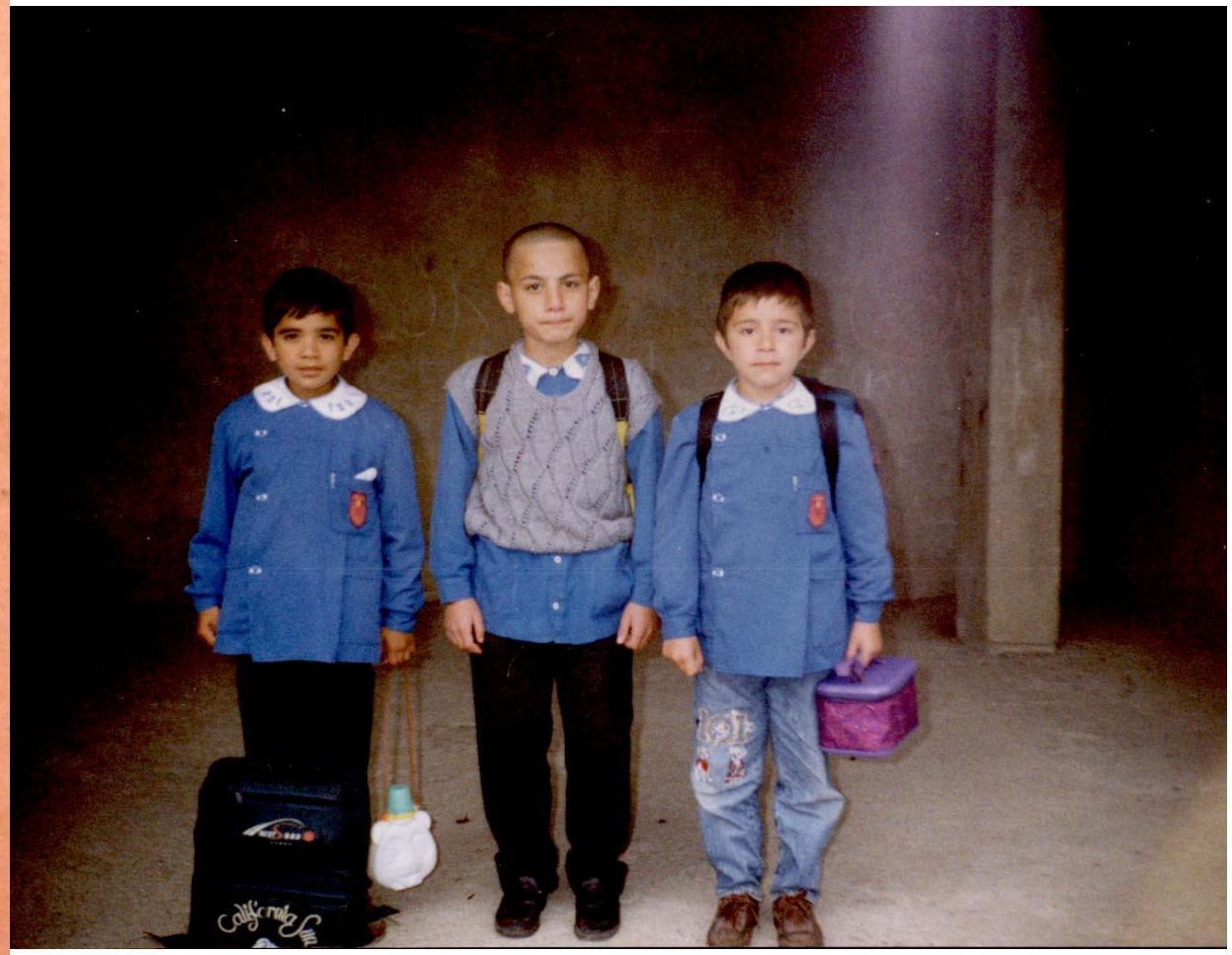
The State of Cystic Fibrosis in Turkey

In Turkey the infant mortality rate is still very high (59/1000). Respiratory infections and diarrhea are important causes of infant mortality (13%, and 9%, respectively) (1). There are no reliable statistical data available for the incidence of CF in our country. Probably some infant deaths which are listed under respiratory and gastrointestinal problems are due to CF, because of lack of diagnosis.

marriages are very common (21%) (2). Therefore we see a large number of inherited metabolic diseases, for instance phenylketonuria (PKU). The mean incidence of PKU is 1:10 000 all over the world, whereas its incidence in Turkey is 1:6 000 according to our studies (3). CF leads to mortality, but PKU to a sequal, mental retardation. We believe that because of the lack of diagnosis we cannot reach patients with CF in our country, but patients with PKU who survive with mental retardation are more easily diagnosed since newborn screening is not organized nationwide as yet.



Development of Pediatric Pulmonology

- 1997- Istanbul University Department of Pediatric Pulmonology

  2002- Pediatric Pulmonology Society
- 2011 Pediatric Pulmonology Board Certification
- 2012 CF Patient Organization

Am J Med Genet. 2002 Dec 1;113(3):250-7.

Highest heterogeneity for cystic fibrosis: 36 mutations account for 75% of all CF chromosomes in Turkish patients.

Kilinç MO[1], Ninis VN, Dağli E, Demirkol M, Ozkinay F, Arikan Z, Coğulu O, Hüner G, Karakoç F, Tolun A.

Development of Pediatric Pulmonology

2011 Pediatric Pulmonology Board Certification

Uzmanlık Tescil No. : 90808
Tıp Tescil No. : 29953

T.C.
SAĞLIK BAKANLIĞI

UZMANLIK BELGESİ

Hacettepe Üniversitesi Tıp Fakültesi'nden 30/06/1980

tarihinde ***80-AA-130*** *sayılı diploma ile mezun olan* ***İstanbul-1957*** *doğumlu*

Elif Dağlı

1219 sayılı Tababet ve Şuabatı Sanatlarının Tarzı İcrasına Dair Kanun ve ilgili uzmanlık eğitimi mevzuatı hükümlerine göre,

Çocuk Göğüs Hastalıkları

alanında yaptığı çalışmaların

Tıpta Uzmanlık Kurulu tarafından değerlendirilmesi sonucunda ***25/01/2011*** *tarihinde* ***141*** *sayılı karar ile uzman unvanını kazanmıştır.*

Development of Pediatric Pulmonology

2012 CF Patient Organization

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YARDIMLAŞMA VE DAYANIŞMA
DERNEĞİ


Foto Uasin
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