39 "Adult onset CF": previous misdiagnoses

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Background: Now that the improved treatment of CF patients means that most are surviving until adulthood, there is an increased awareness of the condition amongst the adult medical community such that more patient are being diagnosed with the condition as adults (>16 years of age). All these patients were misdiagnosed with another disease prior to this correct diagnosis: we were interested in looking at this further.
Aims: We reviewed the last 30 patients referred to our unit with a new (adult) diagnosis of CF, looking at mode of referral, clinical features, and previous diagnoses. Also see adjacent abstract.
Results: The mean age at diagnosis was 29 years (range 16–51), 17 (57%) were female. Twenty (67%) were referred by a chest physician and 5 (17%) by general practitioners, the remainder by paediatricians. All had recurrent chest symptoms, and 4 (13%) also had bowel dysfunction. Prior diagnoses included asthma (20%), bronchiectasis (30%), ABPA (3%), sinusitis (7%), recurrent respiratory infection (7%), unexplained cough (7%), bronchiectasis and coeliac disease (3%), and asthma and bronchiectasis (23%). The mean FEV1 at diagnosis was 63% predicted (29–120) and mean BMI 24 (14–35). Only 11 patients (37%) had a previous hospital admission and only 10 (33%) had previously received intravenous antibiotics. As regards genotype, 1 was ΔF508 homozygous, 15 ΔF508/-, 6 ΔF508/R117H, 2 ΔF508/R117G, and the remainder a mixture of ΔF508, ΔR553X, S1235R, D1152H, and G551D.
Conclusion: A significant number of CF patients are now diagnosed in adult life. All the cases reviewed here had previously been misdiagnosed with another respiratory condition. Adult physicians should consider the diagnosis of CF in patients with unexplained respiratory and bowel symptoms.

40 Prognosis and clinical features of adult diagnosis CF

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Introduction: Although most cases of CF are diagnosed in early childhood, improving treatment means that nearly all are surviving to adulthood such that in the UK, there are now more adults than children with the condition. This has raised the profile of CF with adult physicians, who are now re-analysing patients with unusual chest disease from a CF perspective. To look at this further we reviewed the clinical features of adult patients referred to our unit with a new diagnosis of CF.
Method: We looked at the clinical features at presentation and subsequent outcome in the last 30 patients referred to our regional unit where the diagnosis of CF had been made in adulthood (>16 years of age). Also see adjacent abstract.
Results: Mean age at diagnosis was 29 years (range 16–51), and most presented with recurrent chest symptoms. The diagnosis was made by genotype (46%), genotype and sweat test (47%), and a combination of genotype, sweat test and nasal potential difference (7%). At diagnosis, the mean FEV1 was 63% predicted (29–120) with a mean BMI of 24 (14–35); 15 patients (50%) grew *Pseudomonas aeruginosa* in their sputum, 9 (30%) *Staphylococcus aureus*, 4 (7%) *Haemophilus influenzae* and 4 (13%) had no growth. At follow up (mean 9 years later [1–23]), 27 patients (90%) were still alive (mean FEV1 60% predicted [21–111], mean BMI 25 [18–28]). Four patients (13%) have developed CF related diabetes mellitus.
Conclusion: Patients diagnosed with CF during adulthood have a better nutritional state, less severe lung disease which deteriorates more slowly, and are less likely to be infected with *Pseudomonas aeruginosa*, factors which are associated with a good prognosis. Adult CF units should be aware of this growing population of late diagnosis patients, who will form a significant proportion of their workload over time.

41 Dehydration: a common clinical feature suggesting the diagnosis of CF in Greece

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Introduction: CF presents with various symptoms from the respiratory, the gastrointestinal system, and also as dehydration and male sterility. Dehydration can develop progressively, usually in infants, or acutely.
Purpose: Our purpose was to investigate the ways of clinical presentation of CF in the Greek population, to point out that dehydration is a major symptom leading to the diagnosis of CF and prove its correlation with the environmental conditions.
Material and Methods: In a large review of the years 1986–2007, we investigated the way of diagnosis of CF the Greek population focusing and further analyzing the cases of dehydration.
Results: In a total number of 491 children (males: 249 (50.7%), females 242 (49.3%) the diagnosis of CF was suggested due to symptoms of the lower respiratory tract:141 (29%), the gastrointestinal system: 152 (31%), dehydration:112 (23%), meconium ileus: 64 (13%), nasal polypus: 13 (2.6%), obstructive azoospermia: 8 (14%). Out of the 112 children with dehydration 55 were males (49%) and 57 females (51). 80 of them were aged less than one year old at the time of diagnosis (71.4%). The cases of dehydration during the warm months of the year were 95 (85%), and especially in July 49 cases were diagnosed (44%). During the especially warm years 1987 and 2007 the frequency of dehydration was 50% and 30% respectively of the total percentage of the cases of CF that were diagnosed.
Conclusions: Dehydration is a common symptom of CF that can contribute to the early diagnosis of the disease before the children start presenting symptoms from the respiratory or the gastrointestinal system. Furthermore, dehydration acquires continuously more importance due to the environmental changes that threaten our planet.

42 Reproductive attitudes of parents of CF child: pregnancy termination vs choice to maintain an affected pregnancy

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This study aimed to report data on CF-affected pregnancies in families having CF child(ren), during the 1989–2006 period, in Brittany (Western France), where CF is frequent (1/2800 live birth).
We described the couples already having an affected child, who required prenatal diagnosis (PD), and chose not to terminate the pregnancy when it was positive, and those who chose not to have PD and gave birth to a CF child.
During the 18 year period, 268 PDs were performed in couples living in Brittany. Of them 195 (72.7%) occurred in parents of CF child(ren) and detected 55 (28.2%) affected fetuses.
The rate of terminations was 92.7% (n = 51), plus 1 miscarriage and 3 births. Continuations were based on deliberate/religious choices, and births occurred in families with classical CF [F508del/F508del, F508del/N1303K (n = 2)]. The mean age of the proband was 10.9 years at time of birth of the second affected child.
Over the period, our NBS program let us know that 7 CF children (ie.3.1% of the screened) were born in families who did not request PDs. Four of them had eldest with classical CF forms (F508del/F508del (n = 3), I1234V/I1234V), meanly aged of 4.4 y. Three others had a mild affected elder (F508del/R347H, W846X/R117C, F508del/G91R), meanly aged of 3.7 y.
This study showed the real decisions of parents of affected child(ren) in Brittany, toward PD request, pregnancy termination (92.7%), birth of another CF child. In order to have a better picture of the reproductive attitudes in CF families, it will be interesting to get data on the family size and the genetic status of each child.
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