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Chapter 1 : Formats and Editions of Cystic fibrosis - current topics. 1 [calendrierdelascience.com]

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Hence even a medical article is, in a sense, something of an autobiography" - John Chalmers Da Costa Selected Papers and Speeches, The author and the Leeds Regional Cystic Fibrosis Service There follows a miscellany on various aspects of cystic fibrosis CF as seen by one person, a general paediatrician from Leeds in the North of England who qualified in medicine in Over the next twenty years or so this eventually evolved into an increasing and ultimately a major involvement with the treatment of people with CF and the development of the Leeds Regional Cystic Fibrosis Centre. The encouragement and support of the late Mr. Teresa Robinson from the early Eighties; both these colleagues, and those who followed them, were absolutely crucial in the development of the CF service and were among the first specialised CF appointments in the UK. On this local note, for a service to flourish it must be perceived to be benefiting the patients. This must have been the case as many families and patients returned year after year, often from great distances, even from as far away as Hong Kong, for an Annual Comprehensive Assessment by the small increasingly expert team at St. Access to information, images and publications via modern electronic communications The major developments in electronic communications in recent years have been timely for an undertaking such as this History. The unprecedented advances in access to previous published work via the Internet, into Medline and PubMed in particular, and the availability and ease of transferring visual images, have presented an opportunity which I hope will add a slightly more human touch to some of the important contributions over the years. These developments have resulted in the decision to publish this account on the web so as to provide easy access for all those interested in the story of cystic fibrosis. The excellent and widely used website developed by Dr Daniel Peckham and his colleagues in Leeds www. Daniel Peckham has kindly agreed to co-author the New Millennium sections as I felt it would be wise to have a CF expert currently involved in a busy CF centre to ensure accuracy, relevance and credibility to the content and comments relating to recent developments. Main sources of information Where abstracts were available for the more recent papers I have endeavoured to extract the relevant message and reduce the number of statistics as, with the modern electronic databases, the reader can obtain these from the abstracts or the originals if necessary. Also most of the articles mentioned have a [PubMed] link, which from around the mid-Seventies, allows access to the full abstract. I have read and made photocopies of many of the early papers from before from the originals in the library of the Royal Society of Medicine RSM where I stayed regularly during my frequent visits from Leeds to London from until when I was first Chair of the Research and Medical Advisory Committee and then from to Chairman of the Cystic Fibrosis Trust. These trips to the RSM library involved many hours in the basement among the old bound journals. I am also particularly grateful to my friend and erstwhile surgical colleague Mr. Archie Crompton for the German translations of some of these older articles. Professor Craig kindly gave me most of the US Year Books of Pediatrics from to the mid - Fifties and these have been an invaluable source of information. The content of the whole of this History is, of necessity, heavily biased towards publications in English, developments in the UK and my own slant on these developments. It is almost certain that many important contributions in German, French and other languages have been overlooked. The various phases of research and understanding Some research since the identification of CF in , although of basic scientific interest, was of little or no immediate relevance to the treatment of people with cystic fibrosis. Unfortunately, and understandably, in the past a significant amount of CF research has fallen into this latter category, particularly when there was no clear idea as to the nature of the basic defect. Fortunately, since the early Eighties and particularly after the identification of the CF gene in , much scientific research is clearly focused on the investigation and correction of the basic defect, by either gene therapy or pharmacological means. These words were written in the heady days soon after the identification of the CF gene in and before the first papers on gene therapy were published from onwards - before the many problems in correcting the basic defect were experienced. Nonetheless, the identification of the CF gene was the definite turning point in the

investigation, understanding and treatment of CF after which there was steady, focused progress in correcting the basic defect by either gene replacement or pharmacological therapy. So cystic fibrosis is truly a disorder of our times. During the lifetime of many of us, including myself, the condition was first clearly described as a specific entity in when I was 6 years old , the heredity aspects were recognised in , the treatment was steadily improved, the gene and its product were identified in and by the first drug, ivacaftor Kalydeco , to modify the effects of one of the genetic mutations GD , became available to treat patients first in the USA; also in the first multidose trial of gene therapy was started in the UK by the Gene Therapy Consortium and completed in One reader referred to the work as a "Paper Trail History" which seems appropriate. To obtain a general overview of the history of CF It would be appropriate to read the introduction at the start of each decade. Also refer to the section entitled "Some previous publications on the history of cystic fibrosis" in the Future section. Also for those with access to a medical library or the appropriate textbook the following two chapters in the Fourth Edition of the textbook Bush A, Bilton D, Hodson M. Cystic Fibrosis published in provide a concise account of the history from early days to the present time - Appendix A. History of cystic fibrosis to The main entries in the sections of the new Millennium, where each year has an individual section, are in alphabetical order of the first author. Most of the images that appear in the main text have not been transferred to the Topics section. The images do not always approximate to the appearance of the author at the time the reference first appeared! When searching for a particular subject it also helpful to look in the appropriate Topic Some publications on CF are designated as the "megapapers" These are publications that have had a major influence on the understanding, diagnosis, treatment or outlook of people with cystic fibrosis see somemegapapers. Some of these articles definitely influenced our treatment of people with CF and others advanced the general understanding of the condition. In conclusion Very many thanks to the numerous colleagues in so many disciplines and many countries who have contributed to our knowledge of CF â€” not least the patients and their families. These people are too numerous to mention by name. Finally, there will be many people and publications that should have been included but have been omitted and I would welcome comments if there are significant omissions; also if there are factual errors or misinterpretations. Comments from people with CF and their families would also be most welcome. Any anecdotes, images or personal recollections which would add interest to this History would be most welcome. The text is revised frequently "on line" so there will be an opportunity for immediate alterations and additions rather than waiting for the next edition. The first ten years of the New Millennium were added in late and with this latest revision we have added to Please do feel free to contact me, Jim Littlewood, at history cfmedicine. Both parents are healthy carriers as are up to 1 in 25 of the population. If untreated, signs of severe intestinal malabsorption, failure to thrive and lower respiratory infections, which are slow or fail to clear, are present from an early age. Mutations of the CFTR gene, of which there are now around described, result in salt and water transport abnormalities across the lining cells of a number of organs leading to viscid secretions which become infected and difficult to clear lungs or damage by obstruction pancreas and liver. Since , CF has changed from a hopeless condition, fatal in infancy and early childhood, to a chronic condition affecting more adults than children.

Chapter 2 : The Laboratorian - Volume 1, Issue 4

Manny the Frenchie Visits a Girl with Cystic Fibrosis. Spread awareness and please share.

The implementation of the courier service has had a difficult road, from being an unsuccessful exceptional item requested in to its funding in In February of this year the Laboratory was awarded approximately 2. There are many advantages to establishing a state wide courier service for specimens collected from Medicaid patients, including: Specimens will be received within 24 hours of pick up. This will ensure that more children are screened in a timely manner, reducing stress and hardship on parents and clients. During fiscal year , the pilot project will include sites, which represent 60 percent of all THSteps and NBS specimens submitted to the Laboratory, scheduled for regular pick-ups. The sites were determined based upon their location in the state, the number of THSteps and Newborn Screening NBS specimens that they routinely submit to the Laboratory Measured by the daily average of total specimens submitted from January to September , and the number of THSteps and NBS specimens that were rejected because they were too old to test upon arrival in the Laboratory. In the next several months, we will be evaluating the courier pilot program, establishing performance measures, and sending out surveys to measure customer satisfaction with the service. We will also be looking for additional funding opportunities that will allow us to expand the program even further and sustain it for the long term. There are a number of people in the Laboratory who have worked hard to ensure the successful implementation of this pilot project. I want to publicly thank them for their contributions. The administration offices of the Health Department stayed in the Capitol from until The Pure Food Commission was established in , but it did not come under the Health Department umbrella until Early diagnosis of cystic fibrosis is a difficult and emotional issue for affected families, but countless studies have shown that early diagnosis in infants is associated with increased life expectancy, reduced incidences of hospitalization, decreased mortality rates, improved nutrition, and better overall development. Cystic fibrosis CF is a complex and life limiting recessive genetic disorder caused by mutations in the cystic fibrosis gene called the transmembrane conductance regulator CFTR. This gene is located on chromosome 7 and ensures the production of CFTR protein, which is responsible for the movement of chloride ions through cell membranes and influences the regulation of other intercellular pathways. The hundreds of mutations associated with CF, and the subsequent deregulation of cellular ion transport, disrupt major functions in multiple organs, including the exocrine pancreas, lungs, sweat glands, and intestines. In the lungs, CF prompts an endless cycle of impaired mucociliary clearance, bronchial obstruction, inflammation, and infection. In the digestive tract, CF disrupts the production and release of pancreatic enzymes, triggers the malabsorption of fats and critical fat-soluble vitamins, and alters intestinal motility. Early detection of cystic fibrosis through newborn screening can provide critical time for dietary and respiratory intervention prior to the infant becoming symptomatic. Though no cure for CF exists, currently available treatments improve the overall quality of life. These treatments have also increased the median life-expectancy from 32 years in to 37 years in Demographically, CF is one of the most common genetic disorders in Caucasians, with an incidence of roughly 1 in live births. The disease also has a fairly high incidence among Hispanics with 1 in 4, to 1 in 8, live births being affected. For African-American and Asian-American populations, the incidence rates are approximately 1 in 15, and 1 in 32,, respectively. Overall about 30, people in the United States and 70, people worldwide have been diagnosed with CF. The high incidence rate for cystic fibrosis, as well as its significant carrier frequency, non-Hispanic Caucasians: In , the screening panel was revised to include 23 CFTR mutations and two reflex tests. The table below highlights the 23 mutations recommended by the ACMG. At the DSHS, newborn screening for cystic fibrosis is a two-tiered screening process. Universally, all screening programs for cystic fibrosis rely on the detection of immunoreactive trypsinogen IRT. Trypsinogen is produced in the pancreas and is a precursor for the enzyme trypsin. High IRT concentrations have demonstrated a loose positive correlation with CF because the condition prevents the secretion of trypsinogen from the pancreas to the intestines, which leads to elevated blood IRT levels. All live births in Texas require a first newborn screen within the first 24 to 48 hours. For infants with a high IRT concentration on the initial screen, the second

newborn screenâ€”submitted at one to two weeks of lifeâ€”will determine if further testing is required. The addition of cystic fibrosis screening required countless hours of work by the laboratory, case management, and information technology staff. Extensive validation of the laboratory tests was conducted on the modified computer system to ensure that all results and processes for CF are accurate. Case management contacts consulting physicians and submitters regarding the CF screening findings and recommends follow-up care and additional testing. The sweat test, which will be performed at an accredited CF Care Center in Texas, is the gold standard for diagnosing cystic fibrosis and those results, along with the information provided by DSHS, will be used to determine a final diagnostic outcome and a possible treatment plan for the infant. Through the joint efforts of CF Care Center teams, primary care physicians, and now the DSHS NBS program, a diagnosis of cystic fibrosis can be determined before an infant begins to experience symptoms. The screening process in place at the DSHS is expected to provide early detection for new cystic fibrosis patients each year. Additionally, the program works to provide critical immunizations for all of its participants. The Clinical Chemistry Laboratory performs testing for a majority of the specimens associated with THSteps medical check-ups and coordinates directly with medical providers participating in the program to provide quality care for all patients. THSteps medical services are provided by medical providers located throughout the State of Texas. Medical providers utilize the THSteps medical check-ups periodicity schedule to determine when laboratory screenings need to be performed for each patient. Laboratory Services provided by the Clinical Chemistry Laboratory and associated disorders.

Chapter 3 : eCysticFibrosis Review

Volume 1. This is an ex-library book and may have the usual library/used-book markings calendrierdelascience.com book has hardback covers. In fair condition, suitable as a study copy.

The Institute for Johns Hopkins Nursing and the American Nurses Credentialing Center do not endorse the use of any commercial products discussed or displayed in conjunction with this educational activity. Credit Designations Physicians eNewsletter: The Johns Hopkins University School of Medicine designates this enduring material for a maximum of 1. Physicians should claim only the credit commensurate with the extent of their participation in the activity. The Johns Hopkins University School of Medicine designates this enduring material for a maximum of 0. Each Newsletter carries a maximum of 1 contact hour, or a total of 6 contact hours for the six newsletters in this program. To obtain contact hours, you must complete this Educational Activity and post-test before December 29, Respiratory Therapists For United States: Visit this page to confirm that your state will accept the CE Credits gained through this program. Visit this page to confirm that your province will accept the CE Credits gained through this program. Intended Audience This activity has been developed for pulmonologists, pediatric pulmonologists, gastroenterologists, pediatricians, infectious disease specialists, respiratory therapists, dieticians, nutritionists, nurses, and physical therapists. There are no fees or prerequisites for this activity. The estimated time to complete this activity is one hour. The estimated time to complete this activity is 30 minutes. Course Format The eCysticFibrosis Review series will consist of a monthly review of journal literature on key, pertinent topics, emailed as either a newsletter or podcast, to clinicians caring for patients with cystic fibrosis. The timely commentary on current research, best practices and clinical management issues is provided by an expert panel of pulmonary specialists. The activities are delivered as 6 bi-monthly newsletters and 6 alternating podcasts. Participants will have up to 2 years to complete the 6 newsletters and 6 podcasts in order to earn CME credit. January 28, Expiration date: The following relationships have been reported for this activity: Noah Lechtzin has reported that he has served as principal investigator for Vertex Pharmaceuticals Incorporated. In addition, he has served as a consultant for Hill Rom. No other planners have indicated that they have any financial interest or relationships with a commercial entity. Grants to investigators at The Johns Hopkins University are negotiated and administered by the institution which receives the grants, typically through the Office of Research Administration. Individual investigators who participate in the sponsored project s are not directly compensated by the sponsor, but may receive salary or other support from the institution to support their effort on the project s. Disclaimer Statement The opinions and recommendations expressed by faculty and other experts whose input is included in this program are their own. This enduring material is produced for educational purposes only. Use of Johns Hopkins University School of Medicine name implies review of educational format design and approach. Please review the complete prescribing information of specific drugs or combination of drugs, including indications, contraindications, warnings and adverse effects before administering pharmacologic therapy to patients. Statement of Need Based on a review of the current literature, including national and regional measures, detailed conversations with expert educators at Johns Hopkins, and a survey of potential program participants, this program will address the following core patient care gaps: Disease-Modifying Therapies With the continuing emergence of new data, clinicians are not familiar with the current evidence correlating CFTR genotype with pulmonary and nutritional phenotypes. Clinicians are not familiar with the current data on emerging disease-modifying therapies for cystic fibrosis.

Chapter 4 : Cystic Fibrosis: Current Topics. Volume 2 - Europe PMC Article - Europe PMC

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Chapter 5 : Introduction to the History of Cystic Fibrosis

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A variety of cystic fibrosis gene therapy approaches based on viral (adenovirus, retrovirus, and adeno-associated virus) and non-viral (liposomes and receptor-mediated endocytosis) routes are.