Cystic fibrosis (CF) is Ireland’s most common life-threatening genetically inherited illness with 1 in 19 people being carriers of the condition (Brennan 2004). Ireland also has the highest incidence of cystic fibrosis in the world, in fact, it is as much as four times more prevalent in Ireland than other EU countries and the USA (Cystic Fibrosis Registry of Ireland 2007).

Cystic fibrosis was once limited to childhood, however, due to advances in diagnostic tools and treatment regimes, people with CF are living well into adulthood. The multidisciplinary team involved in the care of a person with CF is composed of the dietitian, physiotherapist, doctor, with the nurse or CF nurse specialist, playing a central role.

Cystic fibrosis is a chronic condition and often diagnosed very early in childhood, this means a long relationship is formed between the nurse and the child and parents. Because of the chronic multi-system nature of CF, nurses working in a community setting need to have an understanding of the pathophysiology of the illness and its impact on the growing child (Duffield 1996).

Pathophysiology of CF
Cystic fibrosis is an autosomal recessive condition that arises as a result of a genetic mutation on the long arm of chromosome 7. The gene involved encodes the cystic fibrosis transmembrane conductance regulator (CFTR) which regulates the chloride channel of many secretory and absorptive epithelia, particularly in the airway and intestines. The most common genetic mutation causing cystic fibrosis involves the loss of the phenylalanine protein at position 508 on the gene. This lack of the phenylalanine protein is problematic in that it causes disturbances in the ionic transport of sodium and chloride ions across epithelial surfaces. The resultant cellular dehydration increases the viscosity of mucus gland secretions and can lead to obstruction of glandular ducts. The stasis of mucus in the airways provides prime conditions for bacterial growth and the patient with cystic fibrosis will typically experience repeated pulmonary infections particularly due to Pseudomonas pneumoniae (Ganong 2005).

The mucus plugs can cause problems in other areas such as the pancreatic duct causing pancreatic insufficiency. The exocrine function of the pancreas is affected due to fibrosis of the acinar glands. Pancreatic enzymes cannot enter the digestive tract which manifests as malabsorption of fat, protein and fat-soluble vitamins. A child’s failure to thrive is a diagnostic aid. The Cystic Fibrosis Foundation report 2005 concludes that 23% of children with CF are below the 10th percentile weight for their age and sex. CF related diabetes can develop if the endocrine portion of the gland is affected.

Other glands affected in the patient with cystic fibrosis include the sweat glands. Normal volumes of sweat are produced, however, sodium chloride is unable to be absorbed as the sweat passes through the gland. The patient will excrete four times the normal amount of sodium chloride in sweat. Although this is not problematic for the patient, it is a good diagnostic indicator (Lewis et. al. 2000). The child’s mother may often report her baby “tastes salty” and this might well be the first symptom noticed in CF.

Male sufferers of CF are likely to be infertile. This is a result of mucus blocking testicular tubes which occurs early in life. Females can however become pregnant despite the mucus at the neck of the womb being stickier in CF (Cystic Fibrosis Trust 1999).
Clinical review

Laboratory tests and diagnosis
According to the Cystic Fibrosis Foundation, diagnosis is achieved through certain criteria. These include:

- Two sweat tests detecting elevated sodium chloride
- Presence of obstructive pulmonary disease
- Pancreatic insufficiency (or failure to thrive)
- Family history (Genetic testing) (Rosenstein and Cutting 1998)

Other investigations employed to diagnose CF include chest x-rays, stool specimens to detect the absence of pancreatic enzymes, genetic testing to confirm the 508 mutation and sputum cultures to detect the presence of bacteria typical of CF.

Presenting features
Cystic fibrosis is detected in the newborn by the presence of meconium ileus, an intestinal obstruction that is evidenced by x-ray as distended bowel loops. Meconium ileus in the newborn merits a sweat test to be performed and cystic fibrosis is assumed unless proven otherwise (Cystic Fibrosis Association of Ireland). A child with CF will be failing to thrive and have poor respiratory health and CF can be diagnosed in the infant or toddler stage of development.

Screening
Historically, in Ireland, screening occurred in families where there was one child born with cystic fibrosis, following this, all successive children were screened at birth. However, because of its high prevalence in Ireland, there has been a strong consensus amongst the medical community that it was necessary for the introduction of a newborn screening for cystic fibrosis to be implemented. As of July 1st 2011, bloodspot screening for Cystic Fibrosis commenced in Ireland. The screening has been incorporated into the ‘heel-prick’ test, which now screens for six conditions, namely, Phenylketonuria, Congenital Hypothyroidism, Maple Syrup Urine Disease, Classical Galactosaemia, Homosystinuria and now Cystic Fibrosis. Time will tell just how beneficial the introduction of this screening is to people with Cystic Fibrosis in terms of survival advantage, number of hospitalisations required and overall quality of life. However, some studies have shown that newborn screening programmes serve to provide a means for both early detection and intervention which in turn prevents early CF related deaths (Dankert-Roelse and Merelle 2005) and is associated with better survival when compared to delayed diagnosis (Lai et. al. 2005).

CF and respiratory health
The key principles in promoting respiratory health in persons with CF include:

- Awareness
- Education
- Early intervention
- Active management

According to Yankaskas 2001, pulmonary disease is the primary cause of death in 95% of CF, therefore, the nursing care of respiratory complications is important in slowing the decline in respiratory function. Most of the care of CF patients occurs in the home.
Patients and their families need to be aware of the importance of concordance with medications and physiotherapy regimes, signs and symptoms of respiratory infections and lung function decline and also the importance of prompt action.

Educating patients and their families regarding avoiding contact with those who have infections such as influenza and importance of vaccinations is key. Promoting exercise to maintain good respiratory health is essential. The nurse must establish if the patient fully understands the technique when using inhalation therapies and provide teaching and education when appropriate.

Early intervention will improve the treatment outcome. Signs of infection should be noted early and dealt with to prevent further progression of the illness.

Active management occurs when patients present to hospital for either treatment of exacerbations of respiratory infections with antibiotics or for suppressive therapy during remissions when the patient is well. Isolation of immuno-compromised CF patients when admitted to hospital is a priority but not always possible due to bed restrictions. This is because of their susceptibility to infectious pathogens from other patients and poor infection control techniques of staff. Standard precautions must be exercised before and after contact with a CF patient. Sputum samples should be obtained 4 times a year and during each exacerbation (Yankaskas 2001). If it proves difficult to obtain a sample, the nurse should arrange for the physiotherapist to carry out airway clearance techniques with the patient in order to mobilize secretions.

**Nutritional management**

A child with CF can achieve normal growth, be better equipped to fight off infections and improve their lung function if their nutritional needs are met (Brennan 2004, booklet). Malabsorption is common in persons with CF as calories and nutrients are lost in their stool. A family centred approach should be adopted and parents should be educated on the appropriate diet for those with CF. Children spend most of their week in school, hence the school canteen staff should also be aware of the dietary needs of children with CF.

Most children with CF lack the ability to digest their food adequately and pancreatic enzymes must be taken. The most common pancreatic enzyme taken in Ireland is Creon. A multi-disciplinary approach is effective with the dietician determining the correct dose on an individual basis based on stool amount and consistency and the level of abdominal pain. The patient and their family need to be aware of the importance of compliance with these doses and educated regarding how to take them, i.e. with food. Foods with higher fat content and bigger portions require higher doses of the enzyme. The enzymes must be swallowed whole, not mixed into food and taken at the right time.

Nutritional supplements can be used as an extra source of calories, not as a replacement for meals. These include, energy and protein-rich as well as carbohydrate-rich supplements. Vitamin deficiencies can arise due to malabsorption in people with CF. Most will be prescribed vitamin supplements by the dietician.

The nutritional health of the person with CF can be quantified by objective measures such as growth percentile charts and laboratory tests to determine vitamin levels in the blood.

**Psychological care of the patient with CF**

Parents of children with newly diagnosed CF can be in considerable shock and disbelief particularly if the child appears well to them. Reassurance to parents at the diagnosis stage is important and this can be achieved by avoiding bombarding them with information; it is a chronic condition and parents will need time to gain an insight into the condition. Some of the information may be out of date and can frighten parents. Referral to a CF nurse specialist can serve to alleviate some of the anxiety as she will have up-to-date treatment information. Novel treatments are expanding the life expectancy further and further and this will offer great hope to parents.
Reassurance to parents at the diagnosis stage is important and this can be achieved by avoiding bombarding them with information.

Acceptance can be a major factor in the psychological wellbeing of people with CF and their families, particularly as they get older. Casier et al 2008 support the idea that accepting the limitations of a chronic illness and readjusting life goals accordingly can have a positive effect on the psychological wellbeing of adolescents with CF. Peer groups and social networking groups such as Facebook groups can provide a connection between people with CF all over the world which can be influential, particularly at the cusp of entering adolescence.

The literature highlights a particularly vulnerable stage in the person with CF and refers to it as the “transition”. This occurs when the child leaves the paediatric services and enters the adult services. It is important to recognise that these teenagers may have fears arising from the fact the some of their peers may have already died from their illness and this may cause uncertainty for the future for them. Nurses are in a prime position to support and help both patients and families with the responsibility of managing their own care. Teenagers should be empowered to take control of their care and start by seeing the members of the multi disciplinary team on their own without their parents. This positive approach may help them to become independent and autonomous adults.

Pharmacological treatment of CF
People with CF are now living well into adulthood, which can be attributable to earlier diagnosis, a multidisciplinary approach and effective methods of treatment (Abbott and Gee 1998). The three major treatment goals for patients with CF include:

- providing for adequate nutrition with pancreatic enzymes and vitamin supplements
- prevention and treatment of respiratory infections with oral, inhaled, nebulised or intravenous drugs
- chest physiotherapy and the promotion of exercise to improve lung function and airway clearance.

(Beers and Berkow 1999).

Respiratory treatment
The main drugs used in the management of the respiratory complications in CF include

- Bronchodilators to increase air entry
- Anti-inflammatory drugs to slow the decline in lung function
- Antibiotics to eradicate bacterial infections.

Aerolized antibiotics have proven effective in both the eradication of the initial Pseudomonas aeruginosa infection common in CF and also for suppression of the chronic infection (Flume et al 2007). Aerolized (inhaled) treatments are preferable as they allow direct delivery to the infection site, hence smaller doses are needed which reduces the risk of systemic toxicity (Yankaskas and Knowles 2001). The drugs used include alfa dornase (pulmozyme) and inhaled tobramycin.

Alfa dornase acts by fragmenting extra DNA particles found in the mucus of children with CF; this makes the mucus less viscous. The tobramycin is used to treat the source of lung infections while ibuprofen acts as an anti-inflammatory, thus slowing the decline in lung function (Strawhacker and Wellendorf 2004). Tobramycin can be both inhaled, to achieve the desired concentration in the airways, and also intravenously to reach the peripheries that are blocked by the mucus secretions. The inhalation route is the safer option as intravenous therapy must be administered in the hospital which puts the patient with CF at risk from developing a hospital acquired infection. The alfa dornase can cause hoarseness, voice alterations and pharyngitis, however, these are usually self-limiting (Yankaskas and Knowles 2001). The patient must be educated regarding the side effects of the treatments and be aware of the importance of reporting any symptoms in good time.

Gastrointestinal treatment
Enzyme replacement therapy is the method of treating the pancreatic insufficiency commonly seen in people with CF. Pancreatin is the general name given to all pancreatic enzymes used as treatments. Creon is Ireland’s most popular drug for treating this condition. It is taken orally in capsule form but it is available in powder form also. The capsules have an enteric coat which aids digestion and the active drug is released in the small intestine. It mimics the function of the naturally occurring enzymes in a healthy individual. Creon Micro is used in very small infants. This type of treatment is a life-long therapy and children should be empowered to learn how to take their own medication from an early age. Enzymes can be taken with food and can be mixed in small amounts with baby food. However, skin irritation can arise around the mouth in the dribbling area if the enzymes are in direct contact with the skin. The irritation around the nipple can also be problematic for breast feeding mothers Vaseline is used as a barrier cream to prevent this (Tully 2006, booklet).

It is important to note that for the most part CF patients are well and living in the community. They are only required to attend the hospital during exacerbations of their illness, therefore, a large proportion of the care is community based with regular appointments to outpatient clinics. Compliance and concordance with medication regimes should be promoted.

Conclusion
The ultimate aim in the management of CF is to find a cure, however, while we wait we must endeavour to promote research that will improve the current methods of treatment. The fact that therapies such as gene therapy are in the not too distant future offers hope to the many parents who give birth to children with CF, particularly in Ireland. Nurses need to be equipped with the knowledge of caring for those with CF as its prevalence increases, all the while never losing sight of the goal to promote health and keep people with CF out of hospital and living life to the full in their own home with their family.

Sources of information