Cystic Fibrosis

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Cystic Fibrosis
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Overview
Cystic fibrosis (CF) is one of the lethal inherited autosomal recessive disease that causes persistent lung infections and limits the ability to breathe over time (Cystic Fibrosis Foundation, 2018).

Facts about Cystic Fibrosis
Life Expectancy of people with CF is up to late 30s
More than 3,000 people live with CF (1,000 worldwide)
Approximately, 1,000 new cases are diagnosed each year
More than 7% of people with CF are diagnosed in late adulthood
More than half of CF population is 18 and older (Cystic Fibrosis Foundation, 2018).

Lung disease are 99% of CF mortality (Cutting, 2015, p. 45).

Pathophysiology
CF affects the cells that produce mucus, sweat and digestive juices. These secreted fluids are normally thin and slippery. But in people with CF, a defective gene causes the secretions to become sticky and thick. Instead of acting as a lubricant, the secretions plug up tubes, ducts and passageways especially in the lungs and pancreas (Mayo Clinic, 2016).

More is caused by defects in the cystic fibrosis gene, which codes for a protein transmembrane conductance regulator (CFTR) that functions as a chloride channel and is regulated by cyclic adenosine monophosphate (cAMP). Mutations in the CFTR gene result in abnormalities of chloride transport across epithelial cells on mucus surfaces (Sharma, 2018).

The signs and symptoms of cystic fibrosis (CF) vary from person to person (Cystic Fibrosis Foundation, 2018).

Respiratory symptoms
Persistent cough that produces thick mucus
Wheezing
Breathlessness
Exercise intolerance
Recurrent lung infections
Inhaled nasal passages or stuffy nose
Sweating
Rectal prolapse (Mayo Clinic, 2016)

Dietary symptoms
Reduced CFTR protein
More than 75% of people with CF have a reduced CFTR protein expression

Other symptoms
Aerobic Exercise
Oral pancreatic enzymes (Mayo Clinic, 2016)

Management
There is no cure. The goal of treatment includes preventing and controlling lung infections, removing and controlling thick mucus from lungs, preventing or treating blockage in the intestines, providing enough nutrition and maintaining good weight, physical therapy, dietitian, and social worker who will monitor the severity of symptoms, the CF treatment may vary depending on the hospital (NHLBI, 2018, Treatment).

Type of treatments
Cystic fibrosis is a complex disease that requires the dedication of healthcare providers that focus on providing specialized care and disease management. CF patients live depend on combination therapies that help lesions and get rid of the thick mucus that can buildup in the lungs, open the airways or thin the mucus through inhaling medicines, and improve the absorption of vital nutrients by promoting appropriate enzymes supplement capsules (Cystic Fibrosis Foundation, 2018).

However, as Carter, Grey, and Wagener (2017) outlined, it is vital for patients to fully understand their treatment to ensure positive outcomes by creating specialized individual plans. Care team must utilize strategies that increase communication with patients to "establish and document realistic, meaningful, and achievable goals and ensure follow-up on progress and barriers to progress” (p.76).

Finally, despite tremendous achievements in the fight for the care of CF, too many lives are still being lost. And the more one understands the underlying mechanism of CF, the better equipped one is to handle this illness. In this perspective, genetic research with precision medicine offers the key to not only finding treatments that improve quality of life of CF patients but also in the fight for the finding of the cure for this terrible disease.

Table 1
Summary of different classes of CFTR mutations

<table>
<thead>
<tr>
<th>Mutation Class</th>
<th>Nature of defect</th>
<th>Functional consequence</th>
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</thead>
<tbody>
<tr>
<td>CFTR channel opening</td>
<td>Functional but unstable CFTR protein</td>
<td></td>
</tr>
<tr>
<td>CFTR channel gating</td>
<td>Partially defective CFTR protein</td>
<td></td>
</tr>
<tr>
<td>CFTR channel stimulation</td>
<td>Increased CFTR protein</td>
<td></td>
</tr>
<tr>
<td>CFTR channel inhibition</td>
<td>Reduced CFTR protein</td>
<td></td>
</tr>
<tr>
<td>CFTR channel activation</td>
<td>Increased CFTR protein</td>
<td></td>
</tr>
<tr>
<td>CFTR protein processing</td>
<td>Mutated CFTR protein transported at cell surface</td>
<td></td>
</tr>
<tr>
<td>CFTR protein trafficking</td>
<td>Mutated CFTR protein trafficked to cell surface</td>
<td></td>
</tr>
<tr>
<td>CFTR protein degradation</td>
<td>Decreased CFTR protein</td>
<td></td>
</tr>
<tr>
<td>CFTR protein expression</td>
<td>Lower CFTR protein</td>
<td></td>
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</tbody>
</table>

Diagnosis
In addition to clinical symptoms, doctors diagnose CF based on multiple tests including:
- sweat test (determination of chloride level)
- pulmonary function test
- abdominal imaging
- genetic screening
- stool test

References
Sharma, G. D., & Cystic Fibrosis Foundation, Retrieved from https://www.cff.org/What-is-CF/understanding-cf/cystic-fibrosiscare...