Cystic fibrosis: Case study

Case:

A male child was born in 1990. He did not gain weight normally and had frequent, loose,

foul-smelling bowel movements. At four months of age, he developed a cough that

produced phlegm. A sweat test showed elevated chloride levels, which are diagnostic of

cystic fibrosis.

He was referred to a CF center and was treated with pancreatic enzymes, chest

physiotherapy (to clear excessive secretions in the chest), and antibiotics. He was in

fairly good health for a number of years, although he struggled to gain weight and had

several hospital stays for breathing problems. At age 10, he became infected with the

bacterium Pseudomonas aeruginosa. His pulmonary function worsened, and he

required hospital stays lasting up to two weeks once or twice a year.

At age 14, he began taking a drug, dornase alfa, and showed improvement. Three

years later, he began taking nebulized tobramycin, and show

showed further improvement.

When he turned 18, he transitioned to an adult CF program. He continued to have

frequent pulmonary exacerbations requiring repeated courses of intravenous antibiotics.

He developed complications from the antibiotics, including kidney damage and hearing

loss. As an alternative, bacteriophage treatment was applied to successfully tackle

opportunistic infections.

At age 24, he received a lung transplant. He had maintained normal lung function for a

long time but had to take several medications to prevent infection and lung rejection.

Questions to answer in your Report (FIVE):

1. Outline the disease genetic background, including the disease inheritance,

gene mapping and possible mutations (20%);

2. Describe the course of the disease, physiological changes, opportunistic

infections (20%);

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3. Provide information on the antibiotic, bacteriophage-based and supporting

treatment (20%);

4. Outline modern approaches to gene therapy of CF, including gene editing and

those currently in clinical trials (20%).

5. Discuss advantages and disadvantages of the lung transplantation for CF

(20%