Week 1 Initial Discussion

Scenario 2: A mother brings her 6-month-old daughter to the HCP for evaluation of possible colic. The mother says the baby has had many episodes of crying after eating and, despite having a good appetite, is not gaining weight. The mother says the baby's belly "gets all swollen sometimes." The mother says the baby tastes "salty" when the mother kisses the baby. Further work up reveals a diagnosis of cystic fibrosis. The mother relates that her 23-month-old son has had multiple episodes of "chest congestion" and was hospitalized once for pneumonia. The mother wants to know what cystic fibrosis is and she also wants to know if she should have any more children.

Post an explanation of the disease highlighted in the scenario you were provided. Include thefollowing in your explanation:

- The role genetics plays in the disease.
- Why the patient is presenting with the specific symptoms described.
- The physiologic response to the stimulus presented in the scenario and why you thinkthis response occurred.
- The cells that are involved in this process.
- How another characteristic (e.g., gender, genetics) would change your response.

Overview:

Cystic Fibrosis (CF) is a genetic disorder of the exocrine glands caused by mutations in

CFTR, a cAMP-regulated epithelial chloride channel which results in impairment in chloride

transfer across the cell membranes with ensuing chloride and water accumulation in organs and

causes viscous secretions to develop that block ducts and form cysts (Mccance & Huether,

2019).

The role genetics plays in the disease:

Cystic fibrosis is an autosomal recessive disorder that affects 70,000 individuals worldwide. The condition affects primarily those of European descent and males, although cystic fibrosis has been reported in all races and ethnicities (Cohen & Prince, 2012). One CF mutation gene from each parent is passed down to the child, that will leave the child with two mutated