Cystic diabetes, osteoporosis and liver problems. cf is

Psychology, Behaviorism

Cystic Fibrosis (CS) is a condition which results in the lungs anddigestive system being clogged with thick and sticky mucus.

CS is a genetic defectwhich is inherited from both parents. Symptoms of CS include recurring chest infections and frequent coughingdue to CF exacerbation which occurs due to build-up of thick mucus in the airways. And Difficulty putting on weight which is caused by thick secretions blockingpancreatic ducts, which results in reduced number of digestive enzymes in thesmall intestine. People with CF can also develop other conditions, including diabetes, osteoporosisand liver problems.

CF is caused by a faulty gene on chromosome 7 called the cystic fibrosistransmembrane regulator (CFTR). The faulty CFTR gene is inherited from bothparents, this gene affects the movement of chloride ions in and out of thecell, by disruption chloride channels on the cell membrane (NHS, 2016). In this essay I will focus on how research to investigate the behaviourof the healthy human body can help explain what happens in disease specificallyCystic Fibrosis. Cystic Fibrosis is an autosomal recessive disorder, which means twocopies of an abnormal gene must be present in order for the disease or trait todevelop. The gene in question is the CFTRgene which codes for the CFTR protein.

Cystic Fibrosis develops when a mutatedversion of the CFTR gene is inheritedfrom both the mother and the farther. However an individual can be a carrier ofa single faulty CFTR gene withoutdeveloping Cystic Fibrosis. Being a carrier of the faulty CFTR gene means you are more likely to have children that haveCystic Fibrosis, therefore it is important for parents to know the risk

thattheir children may have CF, despite themselves being healthy (Riordan, 1989). The CFTR protein is a channel protein found on the cell membrane in thelungs and pancreas, its purpose is to pump chloride ions into varioussecretions outside the cell, such as mucus.

Those chloride ions draw water intothe secretions, which dilutes and thins out the secretions. The most commonmutation of CFTR is the ? F508 mutation. This means that the 508thamino acid in the CFTR gene which iscalled Phenylalanine is deleted. The CFTR protein with the ? F508 mutation getsmisfolded and cannot travel from the endoplasmic reticulum to the cellmembrane. This results in a lack of chlorine ions in the bodily secretions suchas mucus, which intern results in thick mucus. (National Heart, Lung, and Blood Institute, 2013). Studying healthy cells showsus what a CFTR protein is and what shape it is supposed to have.

It also showsus where it’s located and its function, all this information is very usefulwhen trying to understand what the defect is in people with cystic fibrosis. Aswe compare a healthy cell to the cell of someone with CF we will immediately beable to tell the differences in chloride concentration and the lack of CFTRproteins on the cell membrane. We can also deduce the genetic cause of thisdefect by comparing the genetic code of a healthy person to someone with CF. In early childhood biggestproblem people suffering with CF face is pancreatic insufficiency. This happensbecause thick secretions jam up the pancreatic duct not allowing digestiveenzymes to make it to the intestine. Without those enzymes proteins and lipidscannot be properly digested and absorbed. Other time this leads to poor weightgain.

Eventually the backed updigestive enzymes in the pancreas degrade the cells that line the pancreaticduct causing local inflammation, which can lead to acute pancreatitis and withrepeated episodes chronic pancreatitis. Which causes the development of cystsand fibrosis. And also the destruction of pancreatic tissue can also compromisethe endocrine function of the pancreas causing insulin dependent diabetes. Later in childhood peoplewith cystic fibrosis start to develop lung problems. Normally the ciliatedcells lining the airways of the lungs move mucus up the airway towards thePharynx this is called mucociliary action. This is important because the mucustransports debris and bacteria.

Thicker mucus is harder to transport for thecilia, so the mucociliary action becomes defective. Which means that bacteriais allowed to colonise in the lungs. And this increase in bacterial load causessymptoms such as coughing, fever and decrease in lung function, this is calledCF exacerbation and is treated with antibiotics. It can be even more serious ifthe bacteria become antibiotic resistant (University of Rochester Medical Center, 2017). Chronic bacterial infectionand inflammation can cause bronchiectasis which is permanent damage to theairway wall, this leads to permanent dilation of the bronchi. If theinflammation erodes into a blood vessel there can be hemoptysis or coughing upof blood. Overtime the repeated CF exacerbations can lead to respiratoryfailure, which is the leading cause of death with CF (National Heart, Lung, and Blood Institute, 2013).

The problems that peoplewith CF face shows us how important every little function in the human body is. It shows us that the smallest changes in the

genotype and therefore thephenotype of our bodies can result in massive issues and even death. It isimportant to understand what these defects are in people with CF as it might bepossible to cure or treat the symptoms to prevent severe outcomes. Knowingdefects to look for can also help diagnose CF quicker. For example some countrieshave started screening for CF in newborns, which helps start treatment earlier.

The screen detects a pancreatic enzyme called immunoreactive trypsinogen (IRT)which is released into the blood when there is pancreatic damage from CF. Ifthat test is positive then a sweat test is performed, if high levels ofchloride ions are detected in the sweat CF in confirmed. The reason behind this isunlike in the lungs and pancreas where chloride ions cannot leave the cell whenCFTR isn’t working, in the sweat glands chloride ions cannot enter the body. Resulting in sweat with high chloride levels on the skin.

Aside from diagnosis knowingwhat the differences are in a healthy body and one effected by CF is helpful intreatment. As I have discussed the major issues are problems digesting lipidsand proteins due to blockages in the pancreas and pulmonary issues due tomucus. (Riordan, 1989) As a way to treat theseissues fat soluble vitamins, extra calories and replacement of pancreaticenzymes can be supplemented to help nutrient and aid healthy weight gain. Interms of pulmonary treatment, there’s chest physiotherapy, which loosens themucus in the airways through contact or inhalers. There are also medicationssuch as N-acetylcysteine which breaks disulfide bonds in mucus glycoproteins, and Dornase Alfa, which is a nuclease which cuts up nucleic acid in the mucusto thin it out.

(Riordan, 1989) To conclude it is veryimportant to study the behavior of both the healthy and unhealthy human body. One of the main reasons for this is so that we can compare the differentbehaviors and determine what the problem is and what is causing the problem. Finding this out is the first step in treating or even curing the problem. Ihave also shown that this is also important for diagnoses and the developmentof new screening methods.