Human Biology ATAR – Task 3: Extended Response

Lung diseases and treatments (7.5%)

Name:			
Time allowed: 1 Lessons			
Section	Your Mark	Marks available	Percentage
Section 1: Report		10	18.5%
Section 2: Validation Test		44	81.5%
		54	**100%**

Declaration of Authenticity

I (Student Name) _____ declare that this work is my own and I have not plagiarised from any source.

Signature:

Date:

Lung disease and treatments

You are to choose **one** lung disease from List A and **one** disease from List B to research and find information about the named aspects of each disease. You will then complete an in-class validation assessment on your research without notes.

DISEASES

LIST A	**LIST B**
Chronic bronchitis	Pneumonia
Emphysema	Pleurisy
Cystic fibrosis	Tuberculosis

Check list

- Cause, or main causes
- Symptoms and diagnosis
- Current treatments…how they work and what they do
- Prevention

Write the names of the diseases you have chosen here:

Disease A – Cystic fibrosis

Disease B - Pneumonia

Marks Table

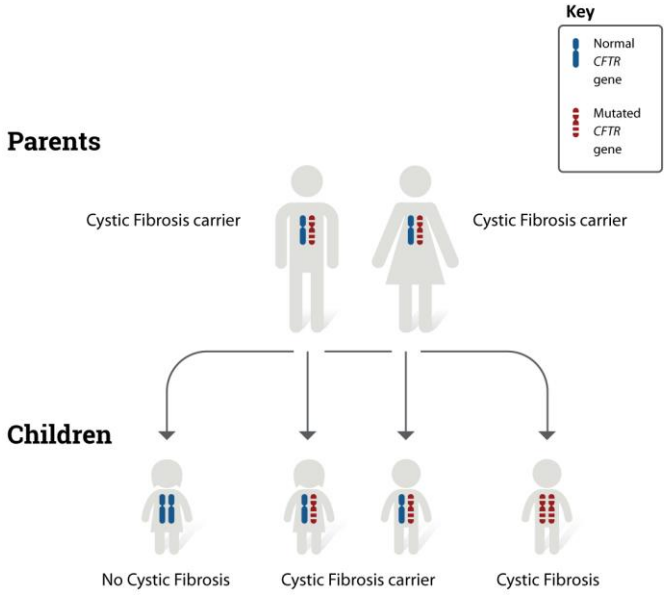
Report	**Cause**	**Symptoms**	**Treatments**	**Prevention**	**Marks**	Your mark
Disease A	1	1	1	1	5	
Disease B	1	1	1	1	5	

This sheet is to be the cover page of your report

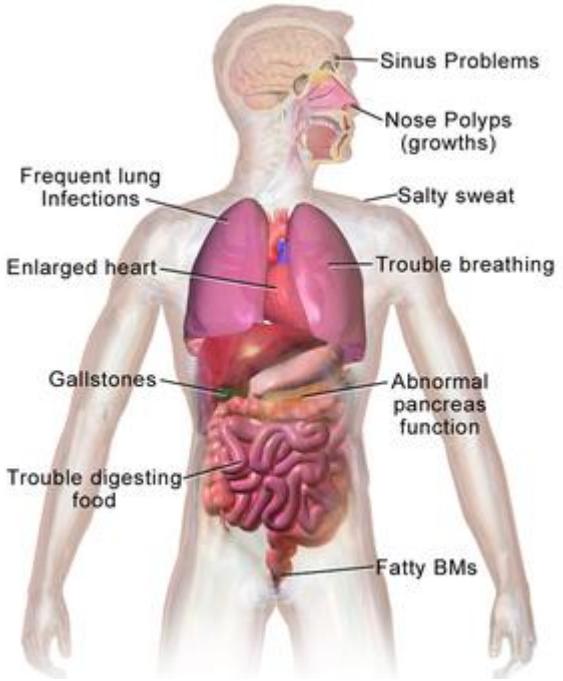
Causes

The CFTR (cystic fibrosis transmembrane conductance regulator) gene is essential because it delivers information to the CFTR protein, which is found in the organs, cardiac cells, and the immune system. When there is a mutation in the CFTR gene, the CFTR protein is unable to function properly, and the cells are unable to move salt and water across cell walls, causing several issues, including having the body producing a thick and sticky mucus, which blocks the lungs and causes complications in the digestive system. The CFTR gene mutation tends to be a processing mutation, which means that a deleted or incorrect amino acid being added to the CFTR protein, preventing it from having the necessary structure to function. Cystic fibrosis is a recessive hereditary condition, which means that the person has inherited two copies of a defective cystic fibrosis gene, one from each parent. For a child to develop cystic fibrosis, both parents must have at least one defective gene and pass it on to the child; otherwise, those who only have one defective trait; one defective gene from one parent and the other is a normal CFTR gene, are called carriers; they do not have cystic fibrosis themselves, but can pass it on to their children if the other parent is also a carrier.

Picture A: Diagram of hereditary chances in cystic fibrosis



Picture B: Common symptoms of cystic fibrosis



Symptoms

Cystic fibrosis effects several systems within the body including respiratory, digestive and the reproductive systems. Many symptoms start early in life (around 6-8 months after birth, whilst other symptoms can go until adult life without detection) and continue to worsen through life or can develop and become more prominent in later years of life, Symptoms are often consisting of sinus issues, enlarged heart, gallstones, abnormal pancreas function, trouble digesting foods (malnourishment, malabsorption of nutrients, unable to gain weight even with a good appetite), recurring chest infections (often pneumonia and bronchitis), frequent wet sounding coughs, diarrhoea, shortness of breath, salty sweat, poor growth/ failure to thrive, difficulty with digestion, internal intestinal blockage or obstructions, infertility, damaged lungs, shorten life span which is usually 44 years, weakened immune system. Due to the nature of the disease people can also develop other conditions such as diabetes, osteoporosis which is weaken bones and liver issues. People with cystic fibrosis may not have all symptoms and can have other systems as each person's symptoms

vary from one another. Often to treat the symptoms can be reduced or prevented with early treatment after detection and medications, with many people with cystic fibrosis needed to be monitored.

Diagnosis

After birth the first screening is a blood test called the “newborn blood spot test” which screens for nine inherited conditions including cystic fibrosis. The test includes pricking the baby’s heel to get several drops of blood which is then placed onto a card and sent to be tested. This test can produce false negatives or not be able to identify cystic fibrosis; only being able to identity 95%-99% of newborns with cystic fibrosis, which is why there is other tests to be done after to confirm the diagnosis. The “sweat test” measures the amount of salt in the sweat, which has been a more reliable test for diagnosing cystic fibrosis than the “newborn blood spot test”. The test includes giving a colourless, and odorless chemical called pilocarpine and an electrical stimulation applied to either the leg or arm to produce sweat from the sweat glands. The sweat is then collected on a piece of filter paper and then sent to a laboratory to be analysed. Those with cystic fibrosis has elevated levels of chloride in sweat compared to those who do not. The test is completed by older children and adults because of its more reliable diagnosis and as a confirmation test for newborns as many newborns cannot produce enough sweat for the test and would need to wait a few weeks.

Treatments

Currently there are no permanent cures for cystic fibrosis but rather only medications which can only ease the symptoms and reduce complications to improve quality of life. The main treatments are to prevent and control infections that are in the lungs, removing and loosening the mucus from/ in the lungs, and treating and preventing intestinal blockages. Medications to use depends greatly on the symptoms presented in the patient however usually is antibodies to treat and prevent lung infections (amoxicillin, clavulanic acid, cefdinir), anti- inflammatory medication to reduce swelling in the airways in lungs, mucus-thinning drugs such as hypertonic saline which helps the person cough up the mucus, acid reducing medication to help pancreatic enzymes to function properly (Creon, pancreaze, xenpep). The new drug called “Trikafta” report helps patients the most, not only easing symptoms but prolonging life. Trikafta being the new breakthrough medicine in cystic fibrosis community, has reportedly improved patients' day to day life most effectively compared to many other medications used by patients. Most medications are tablets which needs to be taken orally whilst some may have the option to be compounded into a powder to be taken for younger children or those who cannot sallow tablets. Many of the treatments must be prescribed by a doctor and dosages are to be advised by a professional.

Prevention

Because cystic fibrosis is an inherited genetic disorder, it cannot be prevented, but there are ways to reduce the risk of passing the condition on to offspring. Carrier testing can help reduce the likelihood of having kids with cystic fibrosis since it detects whether you and your spouse are both carriers of the mutation that causes cystic fibrosis. The procedure requires providing of a blood or saliva sample to a laboratory for analysis. If both parents carry the mutant cystic fibrosis gene, there is a 25% chance the kid will have the condition, or a 50% chance the child will be a carrier but not have the disease. Some couples bear the risk and produce a child "naturally," while others prefer other means such as adoption or utilising an egg or sperm donor who is not a carrier of the illness. If the couple desires a biological child, they can use in vitro fertilisation (IVF), which uses only unaffected embryos to be placed, which means the embryos

must be tested before implantation. Genetic counselling is also done if both parents are carriers as it allows understanding the risks of having a child and allow them to plan and discuss their options.

Causes (no date) *National Heart Lung and Blood Institute*. U.S. Department of Health and Human Services. Available at: https://www.nhlbi.nih.gov/health/cystic-fibrosis/causes#:~:text=What%20causes%20cystic%20fibrosis%3F,instructions%20for%20the%20CFTR%20protein. (Accessed: April 29, 2023).

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Causes (no date) *National Heart Lung and Blood Institute*. U.S. Department of Health and Human Services. Available at: https://www.nhlbi.nih.gov/health/cystic-fibrosis/causes#:~:text=What%20causes%20cystic%20fibrosis%3F,instructions%20for%20the%20CFTR%20protein. (Accessed: April 29, 2023).

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- Prevention

Causes

Pneumonia is a respiratory infection that affects the lungs severely. The lungs in the body have small sacs called alveoli which fills up with oxygen when breathed in, however when someone with pneumonia the alveoli will inflame and fill up with pus and fluids instead which makes breathing difficult, limits the oxygen the person can breathe in and causes fever or chills. Pneumonia can be caused by several reasons which include, bacteria (usually either streptococcus pneumoniae pneumococcus or in children mycoplasma pneumonia), viruses (such as influenza A, and human adenovirus), and recently cases of coronavirus (covid-19) can lead to pneumonia more specially SARS_CoV-2 when contracted leads the virus to attack both lungs caused bilateral interstitial pneumonia. Pneumonia often targets those with weaker immune systems or whilst someone has a weak immune system such as those with HIV, patients having cancer treatments, elderly patients with chronic diseases. More rarely causes of pneumonia have been caused by fungi (pneumocystis jirovecii and aspergillus) which usually only occur in those with weak immune systems,

parasites, aspiration (when food or liquid is breathed into the airways or lungs leading to inflammation and infection), and environmental factors (being exposed to air pollution for example increased risk and can lead to people developing pneumonia).

Symptoms

Symptoms of pneumonia often either rapidly develop or come on extremely slowly, however symptoms usually include; coughing (often producing phlegm or mucus that can be ranging from thick yellow, green or brown), difficulty breathing with breaths being rapid and shallow, also can be shape and feels 'stabbing', rapid heartbeat, fever (over 38 degrees Celsius), sweating or shivering, loss of appetite and chest pains. Other symptoms which are not as common is haemoptysis, which is coughing up blood, blue colour around the mouth on the skin called cyanosis which is caused by the lack of oxygen headaches, fatigue, nausea, abdominal pain, wheezing, joint and muscle pains. In severe cases of pneumonia symptoms include delirium and changes in mental state.

Diagnosis

Before a diagnosis with confirmation tests may be made, the patient must visit a GP (general practitioner). These tests begin with a question about your symptoms, followed by an examination of the chest by tapping it since healthy lungs have a different sounding tap than those with pneumonia, and listening to the chest with a stethoscope for crackling or rattling noises. If the GP believes the pneumonia case is mild, they will likely not refer for an x-ray; however, if they have a reasonable belief, they will recommend an x-ray as the test can show the presence of a pneumonia infection or not, a mucus test (called a sputum test), or blood test, specifically a CBC (complete blood count) to identify if the immune system is fighting an infection, which is identifiable if the level is below a specific number. In rare situations, a pulse oximetry test is performed in which a pulse oximeter is placed to the finger or ear to measure the amount of oxygen in the blood. If the patient is already in the hospital, a blood gas test, in which the blood oxygen levels are measured using a blood sample from the artery, usually the wrist, or a blood culture, which determines whether a germ or bacteria is causing pneumonia and whether it has spread to the blood in the system, is more commonly performed.

Treatments

Viral pneumonia is one of the pneumonia infections which may go away on its own, but it will have extremely high chances the person will be re-infected and become more serious the next time the patient is infected. In mild cases of pneumonia, it can usually be treated at home with getting rest, taking fluids and taking antibiotics specifically amoxicillin as it is usually the most effective is pneumonia cases, due to most cases being bacteria antibiotics are able to kill it if not other treatments should be used. However, for at-risk groups or those with more severe cases of pneumonia other treatments need to ensure such as hospital admission to be able to have oxygen therapy which is oxygen being passed though into a face mask which is worn by the patient to be able to breathe. Due to several causes of pneumonia, there is many ways to treat pneumonia depending greatly on the causes and severity however the most common solutions are taking a medication to aim at the infection (antiviral or antibiotics) the medication will then try to kill the infection if it cannot other ways or medication needs to be taken to get rid of pneumonia and then other medications to reduce symptoms and in rarer cases oxygen therapy or other breathing therapies. Often those who had pneumonia if are healthy and strong can be successfully treated after 2 – 4

weeks whilst for those who have weaker immune systems have higher chances of getting pneumonia again. If pneumonia is left untreated serious complications can follow such as the risk of re-infection, permanent damage to the lungs, bacteria entering the blood stream and effecting other systems in the body.

Prevention

The most effective way to avoid pneumonia is to have vaccinations such as an influenza vaccine as influenza can cause pneumonia and therefore if the virus from influenza is weaken it is less likely to attack and give you pneumonia. The second vaccine which is useful in preventing pneumonia is a “Hib” vaccine (haemophilus influenzae type b) which is a type of bacteria that causes pneumonia commonly and is usually given to children rather than adults. Other ways to avoid pneumonia is keeping the immune system strong which can be by washing your hands/ becoming more hygienic to kill germs, not smoking as smoke weakens the lungs and therefore also defending from infections/ germs, and if having planned surgery do not eat or drink 8 hours before the surgery as this can case aspiration pneumonia because it is more likely to have the food/ liquid fill into your airway if this happens. One of the most important ways to help prevent pneumonia is to be healthy, simply meaning drinking lots of fluids regularly having enough rest, taking in a healthy diet plan and having regular exercise therefore it is less likely for pneumonia be caught and if so, will more likely be easy to get rid of.