The cause of sickle cell anemia biology essay

Science, Biology

SDC in an inherited disease. It can be categorized into two types know as heterozygous sickle anemia and homozygous sickle anemia. A person receive one sickle cell disease gene from one parents and a normal gene from another parent, he will have a condition know as Sickle Cell Trait. When a person receive one sickle cell disease gene from one parents and another sickle cell gene from another parent, he will have a condition know as SDCHow the gene inherited?. During conception, the child gets one set of genes from mother and another set of gene from father. This genes are found on the structure inside the chromosome. When the mothers gene has one gene with sickle and fathers gene has one gene with sickle, then the change of the child being normal will be 25%, 50% chance will be Sickle cell trait and 25% chance of being SDC[1] Please refer Chart 1. In another condition, if one gene with from father and normal gene from mother, the chances are 50% sickle cell trait and 50% normalWhen both allele are effected it now as SDC. This has no heterozygous advantage. Where else, When there is only one allele effected, the person know to have heterozygous advantage. The heterozygote genotype gives a higher fitness compared to homozygote dominant. This happened because heterozygote advantage has single locus refer as overdominance. Overdominance is a genetic condition where the heterozygous individu show a higher finess compared homozygous individu. A classical example is resistnace to malaria infection in sickle cell trait. When pathogen gets into the blood system, the sickle cell stops it in the system untill immune system destroys the pathogen

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The main function of red blood cell is carrying oxygen involved by hemoglobin and protein. Four polypeptides forms the hemoglobin, two alpha chains(each with 141 amino acids) and two beta chain(each with 146 amino acids). This four polypeptides involved in heme-group in transporting oxygen molecules.

The cause of Sickle cell anemia is a " point of mutation`. Its single nucleotide alteration within the beta-hemoglobin polypeptide DNA of gene coding. The sixth DNA triplet, CTC has been changed to CAC (thymine-nitrogenous base is replaced by adenine in the mutan gene).

This mutation results in the change of just a single amino acid within the polypeptide encoded by that gene. Glutamate which normally the sixth amino acid I beta-hemoglobin, with property of an acidic, very polar and hydrophilic side chain. This Glutamate is replaced by valine with property of nonpolar and hydrophobic side chain, in the mutant beta-hemoglobin. This valine at position six forms hydrophobic association with neighboring valine found at position one. This forms the abnormal hemoglobin molecule known as " Hemoglobin S" which is less soluble in water compared to normal " hemoglobin A".

As result of this alteration at the beta chains, When deoxygenated, the hemoglobin molecule will polymerize and " crystallize" becoming long fibris and rigid rod-like structures that will twist into bizarre and jagged shape which also known as sickle shape stucture. When this happened, the loading and unloading of oxygen by the hemoglobin won’t be efficient. Even worse , the cappilarries will might be obstructed by this distorted red blood cells.

Person with sickle cell anemia have a genotype of " HbSHbS" with two mutant beta-hemoglobin. This is know as homozygous for mutated hemoglobin. Individuals with homozygous mutation has no " mutation beneficial". They greatly suffer and usually die early age.

A person inherit one normal and one mutated version of the beta hemoglobin know as heterozygous genotype with " HbAHbS". This group know as " carriers or sickle cell trait" whereby red blood cells generate normal hemoglobin of 60% and mutant hemoglobin of 40%. Sickle cells can be seen in this heterozygous blood, but only 2% - 4% comparing to Homozygous. The heterozygous phenotype have a survival advantage or " mutation beneficial". They generally lead a normal live and heterozygotes (" HbAHbS") survival advantage as they less likely to succumb to malaria. In African many infants with normal hemoglobin die because of cerebral malaria, but there are greater resisitance those with sickle cell trait. Protist Plasmodium falciparum is the deadliest form of malaria which enter the body through mosquito bite. The protist pathogen lodges in human red blood cells, but it decreases the pH of the cells by about 0. 4 pH units, causing about 40% of infected cells to sickle. These deformed red blood cells are sequestered and phagocytized by immune system cells; thus the protist is destroyed along with the sickled cells. Although this does not provide complete protection from malaria, it does lessen the severity of the disease. Sickle cell disease will lead to many clinical complication if it's not managed well. Management of this disease includes management of the anemia, management of pain, Infection management and management of organ damage. Management of anemia. Anemia can because by various condition

in sickle cell disease , a severe anemia cause by homozygosity of sickle cell disease with low hemoglobin. Folate deficiency megaloblastic anemia caused by poor dietary and hyperhemolysis because of infection . To manage, Folate Replacement Theraphy for folate deficiency and blood transfusion for chronic anemia or severe hemolysisManagement of acute and chronic pain episods. Acute pain a condition with unpredictable event with can last from few hours to weeks. A strong Opioids drug given orally to subsidies the pain . Chronic pain defined as last 3 – 6 months, Intervenous (IV) administration with Opioids analgesics used to treat chronic painManagement of infection. As the is absence of spleen in SDC, patient has increased chance of infection to polysaccharide encapsulated bacteria such as S. pneumoniae and H. influenzae. To prevent infection, polyvalent vaccine given every 3-5 years and Haemophilus influenza type b vaccine to be given at age 2, 4 and 6 monthsManagement of organ damage. Types of damages are Central Nervous System which can be in form of Cerebral infraction which managed with exchange transfusion, Cerebral haemorage managed with surgey and seizure disorder managed with anti-epileptic therapy. Opthalimic complication , this involves retinal nevovascularization, retinitis proliferans and erinal haemorhage . This Opthalimic complication managed with photocoagulation(argon or xenon) laser therapy. Respiratory failure involves Acute Chest Syndrome which causes chest pain, hypoxia, fever, dyspnoea anddecreasung hemoglobin level, to manage this arterial blood gases monitoredThe lentiviral gene theraphy is done by introducing genes into individual cells and tissue so that the disease can be treated. The aim Is to create a normal gene by replacing or repairing the defective genes . Main

outcome of this theraphy, the disease ‘ s pathophysiology is corrected . How does the works, target cells are removed from patient and grown it in a culture. The vector a virus example retrovirus, adenovirus which was genetically altered with a therapeutic gene is inserted into virus. The vector then introduced into the culture with patient cells. Patients target cells are now genetically altered with therapeutic gene through the integration of DNA. This cells are reintroduced into the body. Now the genetically altered cells will produce the desired proteins to produce new normal cells or to repair the cells .