GLOSSARY

For Shwachman-Diamond Syndrome

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GENETICS

Chromosome- structure in the nucleus of the cell which contains the genes responsible for heredity. Normal human cells contain twenty-three pairs of chromosomes. One of each pair is inherited separately from a person's mother and fathers

Gene- heredity unit. Each gene carries the genetic code, to the blue print for a specific protein. Each human cell has about 28,000 genes. About 10-12,000 genes are active in any given cell type.

Gene with mutation- a faulty gene, which may result in an inherited disease.

Recessive- a mutation is said to be recessive if an individual inherits two copies of the gene, one from each parent, to develop an inherited disease. Individuals with one mutation and a normal gene will have no clinical symptoms. They are called "carriers".

SBDS- Shwachman-Bodian-Diamond is the name of the faulty gene that leads to most cases of SDS. The protein product from SBDS is thought to help with the manufacture of proteins from the genes that are active in cells.

BLOOD

Hematology- the study of the blood, blood forming cells and the disorders associated with them.

Bone marrow- soft tissue within the bones where all the different blood cells are manufactured.

Stem cells- cells in the marrow that grow and divide to make more stem cells as well as progenitor cells that lead to the platelets, red cells and white cells.

Plasma- a yellowish fluid in the bloodstream which contains various proteins and other substances. Red cells, white cells and platelets are suspended in plasma.

Hematopoiesis- the production of red cells, white cells and platelets by the stem cells in the bone marrow.

Complete blood count (CBC)- number or percent of blood cells, which include white cells, red cells and platelets.

Red blood cells count (RBC)- small round cells that contain hemoglobin. Hemoglobin, which are necessary to carry oxygen throughout the body. Red blood cells account for almost half of your blood volume.

White blood cell count (WBC)- this test measures the number of white blood cells in the blood. WBC's help fight infections. They are also called leukocytes. The most common kinds of white blood cells are lymphocytes and neutrophils.

Anemia- a decrease in red cells which contain hemoglobin. Hemoglobin, which contains iron, is necessary for carrying oxygen to the body's cells.

Hemoglobin (HGB)- the oxygen carrying pigment of the red blood cells; binds with oxygen in the lungs and is carried to the body's cells.

Hematocrit (HCT)- ratio of red blood cells to plasma in the blood, the portion of the blood's total volume that is made up of red blood cells.

Hemorrhage- bleeding from any sire in the body.

Differential count- this is the percent of different types of white blood cells in the body.

Leukopenia- low total white blood cell count.

Neutrophils- (also known as granulocytes)- a mature white blood cell that fights bacteria infection.

Neutropenia- a low absolute neutrophil count (ANC). When the count falls below 1500 per micro liter of blood.

Bands- immature neutrophils. These are usually counted as neutrophils when determining total neutrophils in the blood.

Absolute neutrophil count (ANC)- this is determined by adding the percentage of neutrophils or, polyps in the blood with the percentage of bands in the blood, then multiplying that number by the white blood cells count and multiplying the product by 10. This number represents the number of neutrophils which are available for defending the body against infection.

Cyclic or intermittent neutropenia- when the neutrophil count fluctuates between a normal and a low count. The timing cycle averages about every 21 days, and lasts from 3 to 5 days but can vary.

Severe chronic neutropenia- when the ANC (absolute neutrophil count) is below 500.

Phagocytosis- engulfment and destruction of bacteria or damaged cells by some type of white blood cells, including neutrophils.

Chemotaxis (also known as neutrophil mobility) movement of neutrophils towards a bacterium or an era of tissue damage. Neutrophil must be able to migrate to the particular part of the body to fight off infections.

B-cells- lymphocytes that produce antibodies which help fight infection.

Lymphocytes (T cells or B cells)- cells of the immune system, critical for fighting disease and to help eliminate damaged cells.

Platelets (also know as Thrombocytes)- blood cells that prevent bleeding and bruising. Normal counts range from 150,000 to 400,000. They help to form blood clots.

Thrombocytopenia- a condition in which the number of platelets is less than 100,000 per microliter of blood.

Platelet aggregation- the sticking of platelets to each other to form a clot. This ability can be evaluated by laboratory testing. Abnormal results reflect an increased tendency to bleed (poor clotting), despite a normal platelet count.

Petechia- tiny red dots on the skin due to bleeding under the skin. Usually caused by low platelet counts.

Aplastic Anemia- a rare but extremely serious condition that results from the unexplained failure of the bone marrow to produce blood cells.

Melodysplastic Syndrome- an abnormal development of the cells produced in the bone marrow. This might be an abnormality in shape, size or organization of adult cells. Abnormal proliferation of cells can progress to leukemia.

Leukemia- cancer of the white blood cells with uncontrolled increase in white blood cell count t.

Sepsis- an infection of the blood steam or body tissues. Sepsis can be very seriously s and should be treated immediately.

Blood cultures- these are used to detect the presence of bacteria or fungi in the blood, to identify the type present and guide treatment.

BONE MARROW

Bone Marrow- soft tissue within the bones where all the different blood cells are manufactured.

Storma- The supporting tissue of the bone marrow.

Bone Marrow Aspiration- test in which a liquid sample of the bone marrow is removed by needle aspiration and examined or tested. Results from this test show the appearance of blood cell precursors. Cytogenetic analysis and other special tests require this type of liquid sample.

Bone Marrow Biopsy (BMB)- A test in which a solid core of bone marrow is removed with a biopsy needle. The biopsy is helpful in determining the cellularity of the marrow, among other things.

Cytogenetics- is a branch of genetics that is concerned with the study of the structure and function of the cells, especially the chromosomes.

Cellularity- in the bone marrow, the degree, quality or condition of cells that are present. Evaluation of cellularity aids in determination of whether cells lines are increased or decreased within the marrow.

Colony Stimulating Factor and other Factors (also known as a hematopoietic growth factors or cytokines)- substances produced by the body, which stimulate the production of certain blood cells. Some of these substances has been manufactured synthetically. Examples are granulocyte stimulating factor (G-Csf or neupogen) and various interleukins.

Graft versus Host Disease (GVHD)- the donated bone marrow or peripheral blood stem cells. View the recipient 's body and foreign, and the donated cells/bone marrow attack the body.

Bone Marrow Transplant- a procedure to replace damaged or destroyed bone marrow with healthy bone marrow stem cells.

Telemores- is a region of repetitive nucleotide sequences at each end of a chromosome, which protects the end of the chromosome from deterioration or from fusion with neighboring chromosomes.

Blast cells- Immature cells found in the bone marrow. They are not fully developed and do not carry out any particula

PANCREAS

Pancreas- A large gland that lies behind the stomach. It is made up of two parts, the endocrine and exocrine components.

Endocrine- this component of the pancreas secretes insulin and other hormones, which are necessary to control blood sugar.

Diabetes Mellitis- this occurs when the endocrine portion of the pancreas produces none or insufficient insulin. Insulin is a hormone responsible for controlling blood levels of glucose and for the absorption of glucose in the cells of the body. Glucose is needed for energy.

Hypoglycemia- a condition characterized by abnormally low blood glucose (blood sugar) levels, usual less than 79 mg/dl.

Exocrine- The component of the pancreas that produces several digestive enzymes, including amylase, lipase, protease and trypsin.

Amylase- The digestive pancreatic in sign that breaks down ingested starches.

Lipase- The digestive patriotic enzyme that breaks down digested fats.

Proteases- The pancreatic enzyme (multiple types) that breaks down digesting proteins.

Trypsin- and important pancreatic enzyme which breaks down protein.

Malabsorption- when the body does not absorb nutrients, vitamins and minerals properly. Malabsorption may impair normal growth and development. Failure of the exocrine pancreas or pancreatic insufficiency may cause malabsorption.

Pancreatic dysfunction or exocrine pancreatic dysfunction- evidence of loss of digestive enzyme production in the pancreas.

Pancreatic insufficiency- The pancreas does not produce enough enzymes to digest food.

Pancreatic sufficiency- The pancreas is not normal, low function, but has sufficient function for digestion without the continuing need for pancreatic enzymes with meals.

Steatorrhea- presence of excessive fat in the stool.

Medium chain triglycerides- fats which are most easily absorbed by the body. These can be found in palm oil and coconut oil, for example.

Polyunsaturated fats- fats that are liquid at room temperature.

Fat soluble vitamins- vitamins A, D, E and K; the vitamins that depend on normal digestion to be absorbed properly.

SKELETON

Coxa Vera- a deformity of the hip in which the angle between the neck and the head of the femur, and a shift of the femur (thigh bone) is reduced causing shortening of the leg and a limp.

Genu varus- bowing of the legs at the knee joint.

Genu valgum- knock-knees. The opposite of Genu varus.

Growth plate- this is the hyaline cartilage plate in the metaphysis at each end of the long bone. The plate is found in children and adolescents; in adults, who have stopped growing, the plate is replaced by an epiphyte all line.

Metaphysis- growth plates at the end of the long bones.

Syndactyly- defect in which two or more fingers or toes are joining together.

Thorax- chest.

Tibia- bone of the lower leg, also called shin.

Dysplasia- any abnormality of growth, abnormal size or shape. In Shwachman Diamond syndrome patients, this term may be used to describe abnormalities of the skeleton.

DEXA Scan- dual energy X-ray absorptiometry- a type of one scan that measures bone mineral density.

GROWTH AND NUTRITION

Failure to Thrive- growth failure in an infant or small child. Usually due to inadequate food intake or excessive losses due to malabsorption.

Growth velocity- the rate at which a child grows.

Short Stature- below average height for age, i.e. short, but not malnourished.

Malnutrition- poor nutritional state due to inadequate food intake, a chronic disease or malabsorption. Usually means being underweight.

GENERAL

Bacteria- microorganisms; some can be healthy, such as those that co-habitats and live in your intestine, but they and others can cause serious infections when access other parts of your body, such as blood or kidneys.

Chronic Illness- any long-standing loss or abnormality of bodily function (refers to changes in an individual's body).

Cirrhosis of the liver- extensive scar tissue that forms as the result of damage to,the liver, and may lead to decreased liver function.

Endocardial fibrosis- damage to the lining of the heart and valves.

Hepatomegaly- enlarged liver, without necessarily affecting function.

Hypotonia- Poor muscle tone.

Ichthyosis- A condition in which the skin is dry, rough and scaly.

Immunodeficiency- reduced ability of the body's immune system to fight infections.

Intravenous immunoglobulin (IVIG)- is a plasma product used in the treatment of certain conditions related to the immune system.

Intravenous- injection directly into the vein.

Subcutaneous injection- an injection administered into the subcutis, the .ayer of skin directly below the dermis and epidermis.

Renal tubular dysfunction- eight malfunction of the find tubular part of the kidney, through which water and certain substitute are reabsorbed back into the blood.

Xerophthalmia- I disorder caused by vitamin a deficiency, that can result in severe damage to the cornea.

Fatty Liver Disease (NAFLD)- is the build up of extra fat in the liver cells that is not caused by alcohol. It is seen when more than 5 to 10% of the liver's weight is fat, it is called fatty liver (steatosis).

Antibody titter- the antibody timer is used to determine whether a previous vaccine helped your immune system protect your against a specific disease.