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ASSIGNMENT

The five primary immuno-deficiencies are:

1. PHAGOCYTIC CELL DISORDERS: in this type of disorder, there is an increase incidence of bacterial and fungal infections caused by organisms that are normally nonpathogenic. People with these disorders may also developfungal infections from candida organisms and viral infections from herpes simplex or herpes zoster. These patients experience recurrent cutaneous abscesses chronic eczema, bronchitis, pneumonia, chronic cetitis media and sinusitis.

In one rare type of phagocytic disorder, hyperimmunoglobulinemia E syndrome( formerly known as Job syndrome) white blood cells cannot initiate an inflammatory response to infectious organisms. This result in recurrent bacterial infections of the skin and lungs, abnormalities of the connective tissue, skeleton, vascular system and dentition and extremely elevated levels of igE.

**Assessment and Diagnostic findings**

Diagnosis is based on the history; signs and symptoms and laboratory analysis by the nitro blue tetrazolium reductase test, which indicates the cytocidal activity of the phagocytic cells. A history of recurrent infection and fever including treatment given.

Warning signs of primary immune deficiency disorders are:

* Four or more new ear infection within 1 year
* Two or more serious sinus infections within 1 year
* Two or more months on antibiotics with little effect
* Two or more pneumonias within 1 year
* Failure of an infant to gain weight or grow normally
* Recurrent deep skin or organ abscesses persistent thrush in mouth or fungal infection on skin
* Need for intravenous antibiotics to clear infections
* Two or more deep seated infections including septicemia
* A family history of P.I

**Medical Management**

Attention to infection control practices is important especially with the emergence of multidrug-resistant organism. Although pro phylactic drug treatment effectively prevents some bacterial and fungal infection it must be used with caution because it has been implicated in the emergence of resistant organism. The choice for empiric therapy includes combination regimens and monotherapy.

Specific choices depend on local factors (epidemiology, susceptibility/ resistance patterns, availability, cost). Home and inpatient settings are also available and the selection of setting depends on the patient’s risk category.

Hematopoietic stem cell transplantation (HSCT) another form of cell therapy has proven to be a successful curative modality.

1. B CELL DEFICIENCIES: two types of inherited B-cell deficiencies exist. The first type result from lack of differentiation of B cell precursors into mature B- cells. As a result, plasma cells are absent and the germinal centers from all lymphatic tissues disappear leading to a complete absence of antibody production against invading bacteria, viruses and other pathogens.

This syndrome is called x-linked agammaglobulinemia because all antibodies disappear from the patient’s plasma. B cells in the peripheral blood and igG, igM, igA, igD and igE are low or absent. Infants born with this disorder suffer from severe infections starting soon after birth. Males are at risk for having x linked agammaglobulinemia, if they have an affected male relatives.

Autosomal agammaglobulinemia refers to a rare instance in which normal hypogammaglobulinemia of infancy is prolong.

The second type of B cell deficiency result from a lack of differentiation of B cells into plasma cells. Only diminished antibody production occurs with this disorder.

**Clinical Manifestation**

Infant with x linked agammaglobulinemia usually become symptomatic after the natural loss of maternally transmitted immunglobuline, which occur at about 5 to 6 months of age. Symptoms of recurrent pyogenic infections usually occur by that time. Besides recurrent infection, patients with CVID are at increased risk for autoimmune disease, granulomatous disease and malignancy indicating that CVID is a disease of abnormal immune regulation as well as of immunodeficiency. Patient’s with CVID are susceptible to infections with encapsulated bacteria such as Haemophilus influenza, streptococcus pneumonia and staphylococcus aureus.

**Assessment and Diagnostic Findings**

X- linked agammaglobulinemia may be diagnosed by the marked deficiency or complete absence of all serum immunoglobulins. The diagnosis of CVID is based on the history of repeated bacterial infections. Quantification of B cell activity.

**Medical Management**

Patient with primary phagocytic disorder maybe treated with intravenous immunoglobulin (IVIG) its administration is an essential part of the prevention and treatment of complications of CVID. The use of subcutaneous immunoglobulin has also shown efficacy with easier administration for patients. Antibody replacement therapy is recommended for severe, recurrent infections.

1. T CELL DEFICIENCIES : defects in T cells lead to opportunistic infections most primary T cell immunodeficiencies are genetic in origin. Partial T cell immunodeficiencies constitute a heterogenous cluster of disorders characterized by an incomplete reduction in T cell number or activity. Unlike severe T cell immunodeficiencies, however partial immunodeficiencies are commonly associated with hyper immunedysregulation including auto immune disorder, inflammatory disease and elevated igG production.

Di George syndrome or thymic hypoplasia is an example of a primary T cell immunodeficiency. This rare complex multi system genetic abnormality, which affects multiple organ systems has been mapped to chromosome 10 to 22. The symptoms variation is a result of differences in the amount of genetic material affected.

**Clinical Manifestation**

Infants born with DIGEORGE syndrome have hypoparathyroidism with resultant hypocalcaemia, resistant to stamdard therapy, congenital heart disease, cleft lip and palate, dysmorphic facial features and possibly renal abnormalities. These infants are susceptible to yeast, fungal, protozoan and viral infections and are particularly susceptible to childhood diseases (chicken pox, measles, and rubella) which are usually severe and maybe fatal.

**Assessment and Diagnostic Findings**

Prompt diagnosis is necessary for appropriate management. A comprehensive immunologic laboratory analysis is necessary. Findings in children with DiGeorge syndrome include cardiac and nutritional abnormalities (failure to thrive) and opportunistic skin infections.

**Medical Management**

Patient with T cell deficiency should receive prophylasis for pcp. General care includes management of hypocalcaemia and correction of cardiac abnormalities. Hypocalcaemia is controlled by oral calcium supplementation in conjunction with administration of vitamin D or parathyroid hormone congenital heart disease frequently results in heart failure and these hart patients may require immediate surgical intervention in a tertiary care center. Transplantation of fetal thymus, post natal thymus, or human leukocytes antigen (HLA). Matched bone marrow has been used for permanent reconstitution of T cell immunity.

IVIG maybe used if an antibody deficiency exists. This therapy may also be used to control recurrent infections. T cell function improves with age and often is normal by 5 years of age.

1. COMBINED B CELL AND T CELL DEFICIENCIES: t- cell and B-cell immune deficiencies comprise of heterogenous group of disorders, all characterized by profound impairment in the development or function of the cells or the humoral or both parts of the immune system. A variety of inherited (autosomal recessive and x linked) condition fit this description. This conditions are typified by disruption of the normal communication system of B cells and T cells and impairment of the immune response and they appear early in life.

Ataxis- telangiectasia is an autosomal recessive neurodegenerative disorder characterized by cerebellar ataxia (loss of muscle co-ordination). Telangiectasia (vascular lesions caused by dialated blood vessels and immune deficiency).

Severe combined immunodeficiency disease is a disorder in which both B cell and T cells are missing. Consequently, both cells mediated and humoral functions are affected. SCID is marked by susceptibility to serious fungal, bacteria and viral infections. It refers to a wide variety of congenital and hereditary immunologic defects characterized by early onset of infections defect in both B cell and T cell systems.

Wiskott- Aldrich syndrome a variation of SCID is an inherited immunodeficiency caused by a variety of mutations in the gene encoding the WAS protein. It is characterized by frequent infections, thrombocytopenia with small platelets, eczema and increased risk of auto immune disorders and malignancies.

**Clinical Manifestation**

The onset of ataxis and telangiectasia occurs in the first 4 years of life. Many patients however, remain symptom free for 10 years or longer. As the patient approaches the second decade of life chronic lung disease, cognitive impairment, neurologic symptoms and physical disability become severe. Long term survivors develop progressive deterioration of immunologic and neurologic functions. Some affected have lived until the fifth decade of life.

**Medical Management**

Treatment of ataxia-telangiectasia include early management of infections with antimicrobial therapy, management of chronic lung disease with postural drainage and physical therapy and management of other treatments include transplantation of fetal thymus tissue of HSCT to treat patients with SCID as well as other primary immunodeficiencies. Other treatment options include administration of IVIG or thymus derived factors and thymus gland transplantation.

**Nursing Management**

Many patients require Immuno suppression to ensure engrafment of depleted bone marrow during transplantation procedures. For this reason, nursing care must be meticulous. Appropriate infection control precautions and thorough hand hygiene are essential. Institutional policies and procedures related to protective care must be followed scrupulously until definite evidence demonstrates that precautions are unnecessary. Continual monitoring of the patient’s condition is critical. It Is also imperative that nurses appropriately apply standard precautions (previously known as Universal precautions) which the First-line tools for decreasing transmission of disease; whether from nurse to patient, patient to patient or patient to nurse. Standard precautions are based on the principle that all blood and body fluids, secretions and excretions may contain transmissible infectious agents. Some of the key elements of standard precautions include performing hand hygiene, as mentioned previously, using appropriate personal protective equipment, depending on the expected type of exposure and using safe injection practices.

**Deficiencies of the Complement System.**

The complement system is an integral part of the immune system and deficiencies in normal levels of complement result in increased susceptibility to infectious disease and immune mediated disorders. Improved techniques to identify the individual components of the complement system have led to a steady increase in the number of deficiencies identified.

Hereditary angioneurotic edema results from the deficiency of CI esterase inhibitor, which opposes the release of inflammatory mediators. The clinical picture of thus autosomal dominant disorder includes recurrent attacks of edema formation in the subcutaneous tissue, gastro-intestinal tract and upper airway. Although the disease is mild in childhood and becomes more severe after puberty first episodes have been reported later in life. Food allergy has often been linked to this disorder, although recent evidence has implicated a CL esterase inhibitor deficiency. The fluctuations in hormone levels at the beginning of adolescence, in the premenopausal period, during pregnancy and during the use of oral contraceptives can precipitate an edematous attack that usually disappears after the onset of menopause. Fresh-frozen plasma has been used as well as other treatment options. Paroxysmal nocturnal haemoglobinuria is an acquired clonal stem cell disorder resulting from a somatic mutation in the hematopoietic stem cell . An absent glycosylphosphaticly-linositol (GPI). Encored receptor prevents several proteins from binding to the erythrocyte membrane. These include the complement-regulatory protein-CD55 and CD59- the absence of which result in enhanced compliment-mediated lysis.

Clinical manifestation may be indolent or life threatening. This disorder is characterized by haemoglobinuria that increases during sleep, as well as intravascular hemolysis, cytopenia, infectious, bone marrow, hyperplasia, and a high incidence of life threatening venous thrombosis, which occurs most coomonly in the abdominal and cerebral veins. Severe fatigue, abdominal pain and esophogal spasm may also be present. Lueukopenia, severe infection, and episode crisis are common. severe infection can occur as a result of a plastic bone marrow and splenic thrombosis.

Laboratory diagnosis can include specialized test, such as the sucrose hemolysis test. Ham acid hemolysis and fluorescent activated cell analysis. A coagulation profile is also indicated.