

X-Linked Lymphoproliferative Syndrome

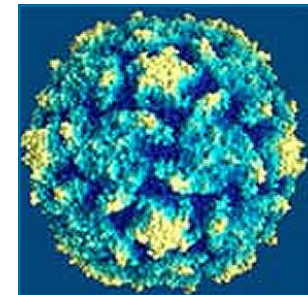
Epstein-Barr Virus

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Introduction

Epstein-Barr (EBV) is also known as infectious mononucleosis, mono, for short, or glandular fever. Epstein-Barr is a type of virus - a member of the herpes family of viruses. EBV is an infectious illness that causes swelling of the lymph nodes. Lymph nodes, containing lymphatic fluid, are part of the immune system, and when swollen they feel like round bumps on your neck, armpits and groin.

The most common age to get glandular fever with symptoms is between the ages of 10 and 35. The incubation period, (the time between being exposed to the virus and getting symptoms), is 33 to 49 days.

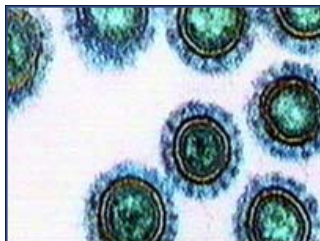
EBV is an infectious illness and can be passed on to family and friends. It is usually passed on through intimate contact between an uninfected person and someone who has the Epstein-Barr virus, but is not showing any symptoms. Only a small number of people (5%) acquire the virus from someone who has developed the full symptoms of the infection contact. The virus is rarely passed on from environmental sources such as coughing and sneezing.

The Epstein-Barr virus is normally spread in the saliva, most commonly through close contact such as kissing.

Symptoms

Symptoms of EBV infection can include:

- swollen, enlarged lymph nodes;
- high fever (temperature above 39C or 102.2F);
- very sore throat;
- swollen tonsils, with a white coating;
- tiredness and lack of energy;
- loss of appetite and weight loss; and
- muscle aches and headache.



EBV virus—courtesy of BBC

The spleen, which is part of the immune system, may swell up noticeably and become painful. This organ is found under the ribs on the left side of your abdomen.

A simple blood test can reveal the level of infection.

EBV and XLP

EBV is a common virus in the normal population. In males with XLP, there is a mutation (mistake) in the XLP gene, *SH2D1A/DSHP/SAP*. This gene helps control the immune response to an EBV infection and codes

for the SAP protein. For males with EBV there is a failure in controlling the proliferation of cytotoxic T cells triggered by the EBV infection which finds a home in the males B cells. As a result, males with XLP who are exposed to the EBV virus can have life-taking threatening symptoms. Patients can experience swollen lymph nodes (glands in the neck or groin), sore throat, fever, and severe hepatitis. After infection with EBV, some patients develop aplastic anaemia (low levels of all types of blood cells) and hypogammaglobulinemia (low levels of antibodies in the bloodstream). Severe symptoms occur because the immune system cannot effectively handle the EBV as in normal individuals and can include severe glandular fever and lymphoma.

Treating EBV

Once a male with XLP is diagnosed with a high level of EBV then the choice treatment is **Rituximab** which removes the antibody-producing white blood cells (B-cells). This has shown to be successful in many XLP affected boys and can be used before and after a bone marrow transplant.

Post Bone Marrow Transplant—CTL's

Bone marrow transplantation is a way to provide healthy T cells to the XLP patient. Although these T cells can control EBV, in the first few months after transplant T cell numbers are very low, while the numbers of EBV-infected B cells may be high. This precarious balance may tip in favour of the B cells and EBV-lymphoma can develop during the recovery period. This problem can be overcome by providing Cytotoxic T Lymphocytes (CTL's) which involves growing up large numbers of T cells from the bone marrow donor that can recognize and kill EBV-infected B cells, in a process that takes about 12 weeks in special laboratories. The T cells are tested to be sure they recognize only EBV and cannot damage normal patient tissues. They are then infused into the patient. T cells are very specific and have few if any side effects and safely provide immunity to EBV.

Definitions

A **Cytotoxic T cell** (also known as a CTL) belongs to a sub-group of T lymphocytes (a type of white blood cell) which are capable of inducing the death of infected cells; they kill cells that are infected with viruses (or other pathogens), or are otherwise damaged or dysfunctional.

Lymphoma is an abnormal tumour mass of tissue produced by an abnormal proliferation of lymphocytes (a type of white blood cell).