Research & Resource Group

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Immunology protocol for patients with Jacobsen syndrome (August, 2017)

Relevant Facts

Jacobsen syndrome (JS) is caused by the deletion of the end of the long arm of chromosome 11 (11q). People with Jacobsen syndrome are at significantly increased risk for having an impaired immune system, i.e. immunodeficiency, which can lead to life-threatening infection, and/or more frequent less serious infections. Between patients the type and severity of immunodeficiency is highly variable. Some patients have life threatening T cell immunodeficiency at birth while other patients develop recurrent infections later in childhood and are found to have other forms of immunodeficiency. Below is a list of the types of immunodeficiency that have been identified in people with JS:

1. Common Variable Immune Deficiency
2. Hypogammaglobinemia
3. T-cell Lymphopenia (including Severe T Cell Lymphopenia)
4. Combined (B and T cell) Immunodeficiency

Based on current research, consideration for immunodeficiency and subsequent immunologic testing is recommended for ALL people with Jacobsen syndrome.

NOTE: Newborn Screening for T cell Lymphopenia has been implemented in many states in the US and other countries around the world, leading to identification of this potentially life threatening condition. This test has identified JS patients with varying degrees of T Cell Lymphopenia. Due to the risk of infection in patients with very low T cells, it is important to have either the Newborn Screening test or a similar assay done on all infants with JS.

We recommend an evaluation of immunity for all infants identified with JS:

- Newborns should have T cells measured soon after birth. This may be achieved via Newborn Screening with a TREC Assay in some states/countries, or through T cell enumeration assays available via commercial laboratories.
- At age 4-6 months: Measure Immunoglobulins (IgG, IgA, IgM)
- At age 12 months: Measure Antibody titers for routine vaccinations such as Tetanus and Diphtheria
- At 2 years: Consider administration of Pneumovax (23 Valent Pneumococcal polysaccharide vaccine) and measurement of post vaccination titers. This helps to identify patients who may lack the ability to mount appropriate immune responses. Using this vaccine has the additional benefit of enhancing protection to Streptococcus pneumoniae in immunocompetent patients.

Older children and adults with JS and a pattern of recurrent infections should be evaluated for the presence of immunodeficiency with similar testing as described above. Consultation with an allergist/immunologist familiar with the evaluation and treatment of patients with immunodeficiency may facilitate diagnosis. Abnormal screening tests, concern for immune deficiency, or recognition of a pattern of recurrent infections should prompt EARLIER EVALUATION, TREATMENT, and REFERRAL TO AN IMMUNOLOGIST!

The following are broader recommendations based on what is known to date regarding immunodeficiency in JS:

- Routine vaccinations should be given unless there is a known T cell deficiency
- Pneumovax-23 at approximately 2 years of age or in older patients with recurrent infections being evaluated for immunodeficiency
- Early treatment of infections with antibiotics
- Repeat immunologic evaluation if infections are recurrent or severe (even if tests were normal the first time)
- SYNAGIS (to prevent RSV infection): STRONGLY consider in children less than 2 years with recurrent respiratory symptoms or any immunologic abnormality, even in the absence of cardiac issues.
- Prophylactic Antibiotics: Some patients with low T cell numbers may benefit from prophylactic antibiotics, especially if the T cell number is felt to increase their risk of opportunistic infections. Other patients with hypogammaglobulinemia and recurrent infections may also benefit from certain antibiotic regimens. Immunologists often prescribe prophylactic antibiotics in an attempt to reduce the frequency and severity of sinopulmonary infections caused by encapsulated organisms.
- Immunoglobulin Replacement: The use of intravenous or subcutaneous immunoglobulin replacement for patients with JS should follow the current recommendations for its use in patients with immunodeficiency.

Additional resources

Identify regional or local immunologists: Jeffrey Modell Foundation (www.info4pi.org)
Information regarding immunodeficiency: Immune Deficiency Foundation (www.primaryimmune.org)
International Patient Organization for Primary Immunodeficiencies: (www.ipopi.org)

Relevant Literature

Dalm VA, Driessen GJ, Barendregt BH, van Hagen PM, van der Burg M. The 11q Terminal Deletion Disorder Jacobsen Syndrome is a Syndromic Primary Immunodeficiency. J Clin Immunol. 2015 Nov;35(8):761-8.

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Disclaimer: This protocol is based on our current understanding of immunodeficiency and Jacobsen syndrome, as of August, 2017. These protocols are subject to change as further information is obtained in the future.