Immunodeficiency in CdLS

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Both physicians and parents have long known that recurrent infections occur commonly in patients with CdLS. Infectious histories collected from 40 patients with CdLS confirmed that recurrent ear infections (59%), pneumonia (47%), and sinusitis (28%) occurred at high frequency. The high infection rate raised suspicion for underlying immunologic abnormalities in CdLS.

At the Children’s Hospital of Philadelphia, we have identified antibody immunodeficiency in a number of patients with CdLS. Antibodies are highly specialized immune proteins which attach to foreign bacteria and viruses, triggering a complex series of events which results in their destruction. Childhood vaccinations are designed to stimulate production of protective antibodies in the body. Patients with defects in antibody function commonly present with recurrent bacterial infections of the upper and lower respiratory tract, such as pneumonia and sinusitis. The identification of multiple patients with immunodeficiency indicates that this may be an under diagnosed and under treated clinical feature in CdLS.

Treatments are available for antibody deficiency and include prophylactic (low dose) daily antibiotics for milder cases and antibody replacement therapy for more severe cases. Given our preliminary findings, patients with CdLS who suffer from severe, recurrent infectious complications should have a formal immune evaluation performed by a qualified clinical immunologist.

This ongoing study is being made possible by a generous grant from the CdLS Foundation’s Small Research Grants Program. Currently, we are continuing to evaluate blood samples from patients with CdLS in order to better understand the mechanisms by which immunodeficiency develops in CdLS and to determine which patients are at greatest risk for this complication.

If your child has immunodeficiency and would like to participate in the study, please contact Dr. Soma Jyonouchi at jyonouchi@email.chop.edu.