

depleted with CD34⁺ selection by using Isolex (Nexell Therapeutics, Irvine, Calif). Initially, he received $10 \times 10^6/\text{kg}$ CD34⁺ cells and $1.9 \times 10^4/\text{kg}$ CD3⁺ cells. He was boosted 7 months later because of neutropenia with $2.2 \times 10^6/\text{kg}$ CD34⁺ cells and $2.5 \times 10^3/\text{kg}$ CD3⁺ cells. There was no graft-versus-host disease (GVHD; see Fig E2 in the Online Repository at www.jacionline.org for engraftment timeline).

A bone marrow aspirate and biopsy, performed 6 months after transplantation, showed normocellular-for-age marrow with granulocytic hypoplasia and increased numbers of maturing lymphocytes (Fig 1, right).

Twenty months after transplantation, our patient had a white blood cell count of 5800 cells/ μL , with an ANC of 928 cells/ μL and an absolute lymphocyte count of 4698 cells/ μL . Cell-surface markers showed a CD3⁺ count of 4252 cells/ μL , a CD3⁺/CD4⁺ count of 2560 cells/ μL , a CD3⁺/CD8⁺ count of 1466 cells/ μL , a CD3⁺/CD16⁺/CD56⁺ count of 287 cells/ μL , a CD45RA⁺ count of 2039 cells/ μL , and a CD45RO⁺ count of 324 cells/ μL . There was normal lymphocyte proliferation in response to mitogens at 20 months after transplantation (see Table E1 in the Online Repository at www.jacionline.org). Intravenous immune globulin was discontinued 20 months after transplantation. The results of hearing studies have been normal.

This case illustrates several important features of RD that differentiate it from other forms of SCID. Our patient presented at 5 days of life, and his initial blood work showed profound leukopenia with severe lymphopenia and neutropenia, which are consistent with previously described cases of RD. He did not have thrombocytopenia or deafness.

As with all forms of SCID, RD is treated with hematopoietic stem cell transplantation. Patients with RD appear to do better with more intensive conditioning before transplantation. We have reviewed the literature specifically mentioning regimens and outcomes for RD (Table I).⁴⁻¹⁰ Patients who received conditioning, specifically with busulfan (16 mg/kg) and cyclophosphamide, had better engraftment and survival than patients who did not. Unrelated cord blood or haploidentical transplantation has been successful. Because RD is so rare and its presentation is earlier than that of other forms of SCID, differentiating RD from other causes of neutropenia and SCID is crucial so that more aggressive conditioning before bone marrow transplantation can be instituted. This case adds to the small number of published reports regarding the treatment and outcomes of RD and further supports the use of myeloablative pretransplantation conditioning for the treatment of RD.

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Disclosure of potential conflict of interest: E. E. Perez has received grant support from the National Institute of Allergy and Infectious Diseases. M. L. Heltzer has received grant support from the Amanda Kanowitz Foundation. The rest of the authors have declared that they have no conflict of interest.

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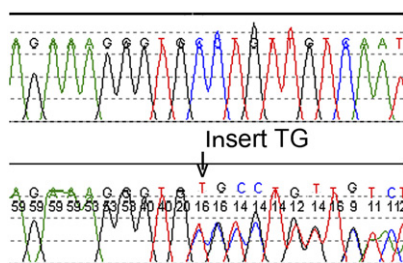
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Available online September 13, 2007.
doi:10.1016/j.jaci.2007.07.026

Complete DiGeorge syndrome associated with CHD7 mutation

To the Editor:

DiGeorge syndrome is an etiologically heterogeneous developmental field defect of the third and fourth pharyngeal pouches, resulting in thymic absence or hypoplasia, cardiac defects, and hypocalcemia. The estimated incidence is 1 in 3000 to 4000 live births.¹ The term *complete DiGeorge* describes patients with the clinical syndrome, profound T-cell deficiency, and lack of proliferative response to mitogens.² It is estimated that 1 of 200 to 250 patients with DiGeorge syndrome has complete DiGeorge syndrome (CDGS). Prompt diagnosis of CDGS is critical because T-cell reconstitution by thymic or bone marrow transplantation is required for patients to survive. Genetic evaluation can be performed to confirm DiGeorge syndrome diagnosis. To date, microdeletions of chromosome 22q11.2 are the only known genetic defects found in CDGS that account for approximately half of patients.² Here we report the first case of the chromodomain-helicase-DNA-binding protein 7 (*CHD7*) mutation in an



A white girl was born via spontaneous vaginal delivery at 35 5/7 weeks gestation because of preterm labor to a 23-year-old mother. Birth weight was 2440 g, and Apgar scores were 1 and 7. The pregnancy was complicated by polyhydramnios and a prenatal diagnosis of esophageal atresia. Maternal depression was treated with sertraline. Born flaccid with gasping respirations, the girl was intubated and later diagnosed with tracheoesophageal fistula with esophageal atresia (type B) and tracheomalacia. Tracheostomy was performed at 2 months of age. Echocardiogram revealed atrial septal defect, ventricular septal defect, and patent ductus arteriosus with pulmonary hypertension. During cardiac repair, a small amount of thymic tissue was noted and excised. She developed hypocalcemia secondary to hypoparathyroidism and was treated with calcium carbonate and vitamin D. Multiple transfusions with irradiated packed red blood cells were given because of intraoperative blood loss. Infectious workup was negative. After preliminary immune

We conclude that the *CHD7* mutation can be associated with CDGS and recommend that subjects with CDGS and CHARGE phenotype as well as children with CDGS who do not meet classical clinical criteria for CHARGE syndrome undergo analysis for genetic defects to include evaluation of *CHD7* gene. In addition, because milder immunodeficiency states have been found in children associated with CHARGE syndrome, immunologic evaluation may be considered in subjects with or without the presence of *CHD7* mutation.

We thank Melissa Dempsey, Genetic Counselor, University of Chicago Genetic Services, Department of Human Genetics, for the use of the sequencing results and Ms Tazim Verjee for the preparation and editing of this article.

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Supported by National Institutes of Health R01 AI47723-04A1 (J.W.S.) and the Eleanor Naylor Dana Charitable Trust (N.T.). Supported in part by the Pediatric Clinical Research Center of All Children's Hospital and the University of South Florida, and by the Maternal and Child Health Bureau, R60 MC 000003-01, Department of Health and Human Services, Health Resources and Services Administration.

Disclosure of potential conflict of interest: The authors have declared that they have no conflict of interest.

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doi:10.1016/j.jaci.2007.08.013

Sensitization to mouse allergen and asthma and asthma morbidity among women in Boston

To the Editor:

Recent studies have shown that mouse allergen is prevalent and potentially important in both urban and suburban

environments, particularly in homes of subjects with asthma sensitized to this allergen.¹⁻⁴ However, little is known about the relation of sensitization to mouse allergen to asthma morbidity in adults with asthma outside the occupational laboratory setting. In this study, we evaluated whether sensitization to mouse allergen is a cause of morbidity in women with asthma from a range of socioeconomic backgrounds from a large metropolitan area in the United States.

This study involved women who were screened for participation in the Epidemiology of Home Allergens and Asthma Study, which has previously been described in detail⁵⁻⁷ (see this article's [Online Repository](http://www.jacionline.org) at www.jacionline.org). This analysis includes 853 women for whom serum IgE to mouse allergen was measured. The study was approved by the Human Research Committee of the Brigham and Women's Hospital. Information on race was determined from the woman's response to questions asking her in which ethnic or racial groups she would classify herself (white, black, Hispanic, Asian, or other) as previously described⁵ (see this article's [Online Repository](http://www.jacionline.org) at www.jacionline.org). Other demographic and socioeconomic variables were obtained from screening questionnaires and have been previously described.⁷

A serum sample drawn at screening was analyzed for total IgE and IgE specific to a panel of allergens using the UNICAP system (Pharmacia, Uppsala, Sweden).⁷ Furthermore, sera were assayed for IgE to mouse urinary allergen (Pharmacia ImmunoCAP e88) and to recombinant Mus m 1 (*Mus musculus*) using the streptavidin technique.⁸ Results for recombinant Mus m 1 showed a strong quantitative correlation with IgE antibody to mouse allergen ($r = 0.92$; $P < .001$). We defined sensitization to each allergen as CAP class level 1 or above (≥ 0.35 IU/mL).

Each woman was asked detailed questions regarding her current and past history of respiratory disease, as previously described.⁹ In the screening questionnaire, women were asked, "Has a doctor ever said that you have asthma?" These women were asked questions regarding asthma morbidity in the past 12 months. A bout of asthma lasting 1 week or more was defined as "prolonged illness," treatment in a hospital emergency room (ER) for asthma was defined as "use of a hospital ER," use of an oral steroid for asthma exacerbation was defined as "use of steroid," and having wheezing or whistling in the absence of a cold (upper respiratory viral infection) was defined as "wheeze without a cold." A positive response to any of the above defined asthma morbidity indicators ("prolonged illness," "use of hospital ER," "use of steroid," and "wheeze without a cold") was defined as "overall asthma morbidity."

The univariable analysis was conducted using χ^2 for categorical variables and 2-tailed t tests for pairs of continuous and categorical variables. Multivariable models were created by using stepwise logistic regression while adjusting for potential confounders and examining interactions. For the interaction between mouse allergen sensitization and race, we dichotomized the data into those who were white and non-Hispanic versus black or Hispanic. The interaction between race and asthma and asthma morbidity was