**GASTROINTESTINAL INVOLVEMENT IN CHRONIC GRANULOMATOUS DISEASE**


**Purpose of the Study.** To evaluate the clinical presentation, prevalence, and consequences of gastrointestinal (GI) involvement in patients with chronic granulomatous disease (CGD).

**Study Population.** A registry of 140 patients with CGD (67% X-linked) maintained at the National Institutes of Health.

**Methods.** This was a retrospective review of records from 1988–2002. GI involvement was defined as abdominal pain, diarrhea, constipation, obstruction or fistulas, and involvement of the esophagus, stomach, or bowel confirmed by endoscopy and/or histology. Other causes of GI involvement were excluded from analysis.

**Results.** Forty-six (33%) patients had documented GI involvement; 44 (96%) were male. Mean age of CGD diagnosis was 2 years (range: birth to 27 years), and median age of GI involvement was 5 years (range: 10 months to 30 years). Thirty-two (70%) patients experienced GI symptoms in the first decade of life, 9 (20%) in the second decade, and 5 (10%) in the third decade. In 8 (17%) patients, GI manifestations preceded the diagnosis of CGD. A high proportion (89%) of those with GI manifestations had X-linked inheritance. All patients experienced severe infections except for 2 kindred, who only experienced GI involvement. Mortality was equal in GI-affected and -unaffected groups and was a result of severe infection. Although all patients experienced abdominal pain, it was the primary presenting complaint in 33% of patients. Other symptoms included diarrhea (39%), nausea and vomiting (24%), and constipation (2%). Obstruction occurred in 35% of patients involving gastric, esophageal, duodenal, and other locations. Despite interferon γ prophylaxis in 89% of GI patients, there seemed to be no protection; 81% of unaffected patients had received similar prophylaxis. After endoscopic confirmation of GI granuloma, successful treatment was initiated by using prednisone (1 mg/kg per day with taper to ~0.25 mg/kg every other day), but 71% experienced relapse. Two patients became hypertensive, and 1 developed cataracts. After bone marrow transplantation, 3 patients experienced remission of GI involvement.

**Conclusions.** GI involvement in CGD is common and recurring, especially in those with X-linked inheritance. Interferon γ prophylaxis does not reduce involvement or affect mortality.

**Reviewer’s Comments.** Although CGD is a rare disorder, the pediatrician must be aware of the classic presentation involving infection of the skin, deep tissues, and bone and complications such as GI granuloma formation. This is especially true in those with X-linked disease. Abdominal pain or abdominal symptoms voiced by a child with CGD must be evaluated thoroughly and, when not infection-related, treated with corticosteroids (in some cases, long-term). Bone marrow transplantation can be effective in inducing remission of the disease including the GI manifestations.

**Health-Related Quality of Life of Children with Primary Immunodeficiency Disease: A Comparison Study**


**Purpose of the Study.** To compare parental perceptions of health-related quality of life (HRQOL) in children with primary immunodeficiency (PI) with children with juvenile idiopathic arthritis (JIA) and healthy children.

**Study Population.** Thirty-six children in each of 3 groups (108 total): those with PI, those with JIA, and those who were healthy. Patients were matched for age, ethnicity, and parental marital status. The age ranged from 4 to 18 years, and 94% were white. All patients with PI received regular infusions of intravenous immunoglobulin. Of the patients with JIA, 77% had either oligoarthritis or polyarthritis. The JIA group had a significantly higher proportion of females.

**Methods.** Parents were interviewed and completed the Child Health Questionnaire-Parental Form 50. Treating physicians completed forms documenting any complications of the underlying disease.

**Results.** In comparison to healthy children, those with PI had significantly lower scores on physical functioning, school and social activities, limitations on parental time and family activities, and parental emotional distress. They were equivalent to the healthy group with respect to overall psychosocial health, daily pain and discomfort, social limitations, self-esteem, mental health, general behavior, and family cohesion. In comparison to the JIA group, children with PI were similar. However, they scored lower than the JIA group with respect to perception of general health and limitations on parental time and family activities. The children with JIA had more bodily pain and discomfort than the children with PI.

**Conclusions.** Children with PI have significant impairment in several measures of HRQOL in comparison to healthy children. These limitations are similar to, and in some cases more severe than, those occurring in another group of chronically ill children, those with JIA.

**Reviewer’s Comments.** This study begins to fill the gap in our understanding of the impact of PI on the quality of life of children and families. Despite some limitations in size and scope, it is clear that HRQOL in children and families with PI is impaired (even when the disease is treated with intravenous immunoglobulin) to a degree that is on a par with other serious chronic disorders that are generally better recognized. Overall, PI is underdiagnosed, to what extent is unknown. This study suggests that not only is there room for improvement in HRQOL aspects of disease management or patient care in those who already have a diagnosis of PI but also implies that there are gains in HRQOL to be made with improved diagnosis of PI.

**Children and Adults with Primary Antibody Deficiencies Gain Quality of Life by Subcutaneous IgG Self-Infusions at Home**


**Purpose of the Study.** To determine the impact of a change from in-hospital infusion of intravenous immunoglobulin (IVIG) to in-home infusion of subcutaneous immunoglobulin (SCIG) on health-related quality of life (HRQOL) and treatment satisfaction.

**Study Population.** Fifty-eight patients between the ages of 2 and 75 years (17 patients <14 years old ["children" for the purposes of this study]; 41 patients ≥14 years old ["adults"] with primary antibody deficiency. Thirty-seven patients were receiving IVIG, and 10 were receiving SCIG.
Health-Related Quality of Life of Children With Primary Immunodeficiency Disease: A Comparison Study

Francisco A. Bonilla

Pediatrics 2005;116;570
DOI: 10.1542/peds.2005-0698

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Pediatrics 2005;116:570
DOI: 10.1542/peds.2005-0698WWW

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