



Adults with Primary Immunodeficiency Diseases

CHAPTER

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Adults with primary immunodeficiency diseases live full lives in the real world. They work, play, marry and have families.

Introduction

Although the first primary immunodeficiency diseases were identified in children, there has been a growing awareness that adults, too, may have a number of primary immunodeficiency diseases. Advances in medicine and earlier diagnosis and treatment of the childhood immunodeficiency diseases have allowed many patients born with primary immunodeficiencies to grow into adulthood. In other cases, many children born with apparently normal immune systems go on to develop a primary immunodeficiency later in adolescence or adulthood.

There are several features of primary immunodeficiency diseases of which a newly diagnosed adult should be aware. In most cases, the well-informed patient, working with attentive medical staff should be able to pursue a career and live a full, active and productive life. This chapter reviews the types of problems that adults with primary immunodeficiencies may develop, discusses how you and your physician can coordinate care, and outlines some of the psychosocial aspects of living as an adult with these disorders.

Common Symptoms

Recurrent infections are the most common problem that patients with primary immunodeficiencies experience. Typically, patients will have recurrent infections in the sinuses (sinusitis) and in the chest (i.e., bronchitis and pneumonia). Early recognition of illness is important to allow timely treatment before infections become severe. Early signs may be as obvious as changes in color or consistency of drainage from the nose or changes in sputum coughed up from the chest, or as subtle as easier fatigability or a shortened temper.

In addition to recurrent respiratory infections, diarrhea is a common symptom that antibody deficient patients may experience. The diarrhea may be caused by a variety of infections or even by an overgrowth of the “normal bacteria” that live in the gastrointestinal tract. Either of the above events results in decreased absorption of important nutrients required for normal body function. *Giardia* is one of the more common protozoal intestinal infections that can cause diarrhea. Patients with compromised immune systems are uniquely susceptible to *Giardia* which can be treated easily with oral medication.

Also, it is not unusual for adults with a primary immunodeficiency to have chronically red eyes, a condition known as “conjunctivitis.” In many patients, if the immunodeficiency can be treated with immunoglobulin, the conjunctivitis often improves, although additional antibiotics are sometimes needed.

Some patients also experience arthritis-like symptoms or other symptoms seen in patients with “autoimmune” diseases. These conditions are covered in specific chapters in this handbook (see chapters titled *Selective IgA Deficiency* and *Common Variable Immunodeficiency*).

It is important that patients be familiar with the common symptoms that accompany their particular diagnosis, so that appropriate care can be sought. Most physicians who provide care to patients with primary immunodeficiencies know that these patients may require frequent antibiotics. Also, these antibiotics often need to be given earlier in the course of an illness and for longer periods of time than in people with intact immune systems.

General Care

It is important for any patient with a primary immunodeficiency to understand as much as he or she can about the workings of the immune system. Knowing when to involve medical professionals may mean staying healthier longer. To help maintain good health, there are things that patients can do in their everyday lives. In particular, good nutrition is of great importance. A balanced diet is essential for normal growth, development, body repair and maintenance, and especially important in preventing and fighting off disease. The general principles of good hygiene are also critical. Simple things like washing hands before meals and after using the restroom go a long way to prevent illness and should become routine habits. Most viruses, including the ones responsible for the common cold, are spread by unwashed hands. Any cuts or scrapes on the skin should be cleansed completely and any

unusual redness or drainage should be reported to a physician so further treatment can be initiated promptly. Dental hygiene and regular dental check-ups are essential since some patients are more prone to tooth decay and gum diseases. Regular exercise helps to maintain optimal function of the body and is also a good means of stress relief for the mind.

Specific treatment for the primary immunodeficiency disease should be coordinated between the patient and the healthcare team members. Each adult should do everything possible to foster good communication between themselves and their healthcare providers. Patients are the only people who can honestly inform their physician about how they feel. They are the ones who have experienced and know which treatments have truly been of benefit and which had not.

The Newly Diagnosed Adult Patient

Some people who have been recently diagnosed have felt unwell for years, without any answer as to what was causing their illnesses and problems. In some cases, a diagnosis can actually be a relief for the patient by finally providing that answer. However, at the same time, the newly diagnosed adult patient must face questions and problems that have already

been faced by children who have grown up with these disorders. Feelings of self-pity and fear are quite normal, but must be identified and addressed promptly. Above all, it is important to realize that you are still the same person, only now you must come to terms with your diagnosis and treatment decisions to create a normal life for yourself.

Self Education

Self education is the key to caring for one's own health. The more an individual understands about his or her primary immunodeficiency disorder, the more confident that person will feel, thus, making treatment decisions easier. Whether they grew up with the disorder or were recently diagnosed as adults, patients must ask questions, obtain educational materials and understand the realities of their deficiency. Most importantly, patients should read about their disorder and become informed, getting involved with their own care. This can help produce a feeling of independence and control over their life. One way to begin this process is to seek out healthcare professionals who specialize in these disorders. Physicians who have little interaction with patients with primary immunodeficiency

diseases may either overestimate the difficulties or underestimate the need for complete evaluation. It is important to ask as many questions as you can of a specialist. No question about your disorder is too trivial. New methods of investigating and treating these illnesses are being developed each year, and it is in the patient's best interest, regardless of age, to find out what these are. Another way to gather knowledge is through contact with other individuals who live with primary immunodeficiency diseases. Immune Deficiency Foundation can put you in touch with another patient through its peer contact program. Talking to other patients and families is often helpful, and any feelings of isolation you may be experiencing can be dispelled.

Advanced Education

A major goal of adults is to be self-sufficient and the importance of receiving an education to achieve this is hard to overestimate. It is important for an individual with a chronic illness or medical condition to obtain a job with good health insurance and a position with enough flexibility to allow appropriate medical attention when necessary. Advanced training and education provide a greater range of choices and flexibility for anyone, and particularly for a person with a preexisting health condition. For young adults who leave home to go to college or other training facility and are on their own, a potential problem is the tendency to “downplay their illness” or fail to disclose their medical history to the school. It is hard for a college or school infirmary to care

appropriately for a student who has not informed the school of his/her specific diagnosis. A primary immunodeficient individual may need antibiotics more often, or sooner in an illness, than another student. For a student with a more serious deficiency, school infirmaries may not be an ideal place to receive care. One way to manage this problem is for the parent or student to find out in advance a local physician with experience in treating patients with a primary immunodeficiency before school starts. Copying records including the patient diary can be of tremendous help in maintaining continuity of care. Should more complicated problems arise, appropriate arrangements can be made. A referral from your current physician may help.

Employment

Adult patients, in choosing a job or career, must think in terms of ones that are suitable for their condition. Depending on the nature of your condition, you may or may not be limited physically. However, there may be complications that need to be fully considered. Factors like time and stress and how they affect your condition and treatment, cannot be ignored. In seeking employment, be aware that there are laws against discriminating against an applicant based on a chronic health condition. However, that does not mean that the laws are easy to enforce. You may want to familiarize yourself with the wording of the laws. For many patients, the health insurance coverage associated with employment is the most problematic. Small employers, for instance,

may not be able to cover you, so perhaps larger corporations and government jobs should be considered while considering careers. New Health Insurance Portability and Accountability Act of 1996 (HIPAA) legislation has improved the ability to transfer insurance coverage from job to job once you are insured (see chapter titled *Health Insurance*). Most patients with primary immunodeficiency disorders work in a variety of jobs. The Family Medical Leave Act (FMLA) also ensures continued employment in the face of prolonged work absences due to illness. Disability in this population is uncommon and usually results from complications of illness and not the primary immunodeficiency itself.

Home Care

Adult patients must learn to fit their treatment into their school and working lives. No longer are patients limited to long cumbersome treatments. Choices mentioned previously including homecare for intravenous immunoglobulin and subcutaneous immunoglobulin administration allow flexibility, minimizing the impact on normal daily living. Home healthcare services permit treatment in your own home environment. This is particularly useful in avoiding missed time from work. You will want to discuss with your physician these treatment

options and ensure that your insurance will cover home healthcare. In many cases, your physician can provide you with the names of a number of home healthcare agencies in your area. Some adult patients who need infusions of intravenous immunoglobulin can learn to give their own infusions. This can be less expensive and more convenient for the working person. In other cases, a nurse who is employed by a home healthcare company can deliver all of the home care.

Health Insurance

Health insurance is an issue that all people with a primary immunodeficiency disorder must face (see chapter titled *Health Insurance*). Decisions regarding school or employment may be affected by insurance coverage. This cannot be taken lightly by anyone with a pre-existing condition. If you allow your insurance to lapse or do not

look into the options that exist before coverage terminates, your ability to qualify for insurance may be seriously jeopardized. It is important for an engaged or married couple to also face the issue of health insurance realistically and understand its importance in career decisions.

Dating, Marriage and Children

Some people may find it difficult to discuss their disorders with their regular friends, and particularly with significant others. It often depends on an individual's own personality as to how much they want to explain, and when they feel comfortable discussing their medical condition. When you do discuss your disorder with your partner, be sure that you make it clear that you have a primary immunodeficiency disease, and that it is not contagious. Often when a patient becomes seriously involved with another person, it may be helpful to have that person accompany them on a visit to the immunologist to better understand the disorder. When a couple is considering marriage, it is important for both to understand the genetic implications of the disorder, and whether it could be passed onto children or grandchildren. Your immunologist or genetic counselors can accurately answer these questions. You may wish to refer to the chapter titled *Inheritance* in this handbook.

Emotional Strains

An adult with a primary immunodeficiency disease has all of the medical problems that a child would have, and yet by the definition of adulthood, is supposed to be responsible for his or her life, career, financial planning, and the future of his or her children. Obviously, this can bring enormous stress into a family. For the adult who has recently been diagnosed, there may be feelings of confusion, self pity and, above all, fear. These thoughts and feelings are normal. The positive side of having a diagnosis is that the uncertainty is over, and you can be on your way to understanding your illness. However, emotional difficulties may arise. It is difficult to have a chronic illness and to be susceptible to repeated or recurring infections in addition to other medical ailments.

Dating, Marriage and Children continued

One of the difficulties associated with primary immunodeficiency disease is the unpredictability of manifestations such as infections. This can place pressures on oneself, family and friends. In addition, the possibility of unexpected absences from work, last minute changes of social activities, or even hospitalizations may cause added tension. Emotional and social problems caused by primary immunodeficiencies are just as important as physical problems and should be discussed with your physician. Sometimes just airing your fears can have a therapeutic effect. This is where an informed friend can be invaluable. True adult friends know when to help and when to motivate you to help yourself.

Another often unspoken stress in families in which the primary immunodeficiency is inherited, may be feelings of guilt on the part of the parent who has passed the defect onto a child. Again, the best way to deal with these feelings is to discuss them

with your family, your physician and your genetic counselor. Remember that this is out of your control, and you have also passed on a number of extremely good qualities. Children with a parent diagnosed with a primary immunodeficiency may themselves have a fear of becoming ill when they are older. In most cases the fear is unfounded and can be dispelled with the proper information and testing.

There are a variety of ways to help keep your frustrations and anxieties to a minimum. You may simply require some time to discuss these feelings with a spouse, understanding friend, clergy or healthcare professional. A number of patients are helped by meeting with others in a support group setting. For many patients, learning as much as possible about an illness is the best way to guard against confusion about the illness itself. Understanding one's own primary immunodeficiency can lead to taking control of one's own life.

Summary

Adults with primary immunodeficiency live in the real world. They work, play, marry and have families like other people. There is no reason why their primary immunodeficiencies should alter this. However, they must be aware of their condition, and use common sense in recognizing symptoms and treating infections. These adult patients must make sure they have access to trained specialists who understand their disorders and are aware of the most recent developments in treatment. They must be careful and informed about obtaining and keeping health insurance coverage and about the laws and regulations that govern insurance. Education and awareness are keys to helping adults with a primary immunodeficiency make good choices and realize their potential.