EDITORIAL

IMMUNODEFICIENCIES: THE CLINICIAN IS NOT ALONE

In all its innumerable chapters, medicine has made substantial advances in the last few years, but perhaps the most spectacular strides forward have occurred in the field of genetics, culminating with the development of the human genome¹, which will help to elucidate the etiology of a whole multiplicity of diseases. In many of these diseases, the causal role of gene-related factors, whether inherited or mutated, is unknown². Therefore, the relationship between genetics and multiple fields of medicine is obvious, and especially so with immunology, including diseases due to hypersensitivity, that is, allergic diseases.

The field of immunology has also shown considerable advances, passing from knowledge of only two lymphocyte lines (B and T) and the role of a few other cells, whose interaction through cytokines was poorly understood, to the current development of analytical technology, which has provided greater insight into the mechanisms through which the body defends itself against pathogenic agents and potentially harmful substances, such as allergens³⁻⁶. Nevertheless, since progress is unstoppable, current knowledge will probably become obsolete and oblige us to change our way of understanding the pathogenesis of diseases of the immune system.

Regarding the pathology of immunity, and especially that of the primary specific immunodeficiencies, new clinical entities have been described since the identification of new lymphocyte subclasses (CD3, CD4, CD8, Th1, Th2, Th3) and better knowledge of cell metabolism and of the elements participating in the interaction between the cells involved in organic defense (cytokines), the numerous cell receptors or histocompatibility antigens.

The first classifications of primary immunodeficiencies included a limited number of processes, most of which were included in B lymphocyte deficiencies, affecting especially the distinct immunoglobulins, while T-lymphocyte or combined deficiencies were given less space^{3,7,8}. With greater understanding of the interaction between T and B lymphocytes, this latter section gradually grew in size^{9,10}. Nevertheless, patients, usually family groups, suffering from frequent and severe infections were often described but commonly used techniques did not allow them to be diagnosed with one of the known syndromes. Improved knowledge of the mechanisms of immunity, due to new study techniques, has led to the identification of a series of new immunodeficiencies, which has greatly added to the chapter on primary specific immunodeficiencies, as reflected in the latest classification¹¹.

Knowledge of specific genes involved in dominant or recessive inheritance, or mutation, increases the complexity of this new classification but also contributes to greater depth of understanding and, probably, to improved treatment of these patients^{10,12}.

The diagnosis of many of these new clinical entities, which are usually due to an alteration of one of the recently identified elements involved in immune response, requires the use of new diagnostic methods, which are not always available in laboratories, due to their low efficiency, the need for specially-trained staff, and occasionally, the difficulties of interpreting the results ¹³⁻¹⁶. Hence the utility of national and international reference laboratories specialized in these techniques; material with elements that cannot be identified without specialized experience and equipment can be sent to these laboratories. The title of this Editorial – The Clinician is not Alone – refers to this collaboration. In this sense, the relationship established between the distinct immunology services and laboratories of Spanish hospitals is a great asset. A list of the techniques that can be performed in each of these hospitals was published in the previous edition of Allergologia e Immunopathologia ¹⁷, which will undoubtedly benefit patients.

F. Muñoz-López

REFERENCES

- 1. International Human Genome Sequencing Consortium. Initial sequencing and analysis of the human genome. Nature. 2001;409:860-921.
- 2. Jiménez-Sánchez G, Childs B, Valle D. Human disease genes. Nature. 2001;409:853-5.
- 3. Fudenberg HH, Suites DP, Wells JV. Manual de Inmunología Clínica. 1.ª ed. 1978. Ed. El Manual Moderno, S.A. México D.F.
- 4. Paul WE. Fundamental Immunology. 1.ª ed.,1984. Raven Press. New York.
- 5. Chaplin DD. Overview of the immune response. J Allergy Clin Immunol. 2003;111:S442-59.
- 6. Janeway CA, Travers P, Walport M, Shlomchik MJ. Inmunobiologia. El sistema inmunitario en condiciones de salud y enfermedad. 2.ª ed, 2003. Ed. Masson. Barcelona.
- 7. Stiehm ER, Fulginiti VA. Immunologic disorders in infants and children. 2nd ed, 1980. W.B. Saunders Co. Philadelphia.
- 8. Chandra RK. Primary and secondary immunodeficiency disorders. 1983. Churchil Livinstone, Edinburg.
- 9. Rosen FS, Cooper MD, Wedgwood RJP. The primary immunodeficiencies. N Engl J Med. 1995;333:431.
- 10. Buckley RH. Primary cellular immunodeficiencies. J Allergy Clin Immunol. 2002;109:747-57.
- 11. Notarangelo L, Casanova J-L, Fisher A, Puck J, Rosen F, Seger R, et al, for the International Union of Immunological Societies Primary Immunodeficiency Diseases Classification Committee. Primary immunodeficiency diseases: an upday. J Allergy Clin Immunol. 2004;114:677-87.
- 12. Fisher A. Human primary immunodeficiency diseases: a perspective. Nature Immunol. 2004;5:23-30.
- 13. Pawliczak R, Shelhamer JH. Application of functional genomics in allergy and clinical immunology. Allergy. 2003;58:973-80.
- 14. Folds JD, Schmitz JL. Clinical and laboratory assessment of immunity. J Allergy Clin Immunol. 2003;111: S702-11.
- 15. Illoh OC. Current applications of flow cytometry in the diagnosis of primary immunodeficiency diseases. Arch Pathol Lab Med. 2004;128:32-1.
- 16. Parisi CAS. Microarray. Arch Alerg Inmunol Clin (Argentina). 2005;36:15-9.
- 17. Español T, Hernández M, Giner MT, Casas C, Gurbindo D, Marco T, et al. Directorio de pruebas diagnósticas de las inmunodeficiencias primarias. Allergol et Immunopathol. 2005;33:157-61.