LICENCIATURA EN NUTRICIÓN

FISIOPATOLOGÍA 1

CUADRO SINÓPTICO: SISTEMA INMUNOLÓGICO

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GRUPO “A”

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FISIOPATOLOGÍA DEL SISTEMA INMUNE

INMUNODEFICIENCIA

Anomalía en uno o más de los componentes del sistema inmunitario, que produce infección por microorganismos invasores o el desarrollo de síndromes neoplásicos.

Deficiencias de la inmunidad humoral (células B)

Agammaglobulinemia ligada al x (ALX): Trastorno hereditario recesivo ligado al sexo, que afecta a uno de cada 250 000 varones. Se deriva de un defecto en el desarrollo temprano de las células B, que determina una disminución intensa en la formación, maduración y sobrevivencia de los linfocitos B maduros. Los varones afectados tienden a contraer infecciones por bacterias encapsuladas, como S. pneumoniae, H. influenzae tipo b, Giardia lamblia, meningococo y distintos enterovirus.

Inmunodeficiencia variable común: afecta a varones y mujeres por igual, no se ha identificado en ella alguna mutación genética específica. Los pacientes con este trastorno pueden presentar insuficiencia del coestimulador inducible de células T (ICOS), insuficiencia de CD19, polimorfismos del gen mutS homólogo 5 de Escherichia coli (MSH5) o insuficiencia del ligando de interacción activador transmembrana y movilizador del calcio (TACI).

Insuficiencia selectiva de inmunoglobulina A: caracterizada por una reducción moderada o intensa de las concentraciones de IgA sérica y secretora. Las personas con insuficiencias graves muchas veces experimentan infecciones respiratorias y gastrointestinales repetidas, y muestran aumento en la incidencia de alergia y otros trastornos autoinmunitarios.

Deficiencias de la inmunidad mediada por (células T)

Síndrome de DiGeorge: Defecto del desarrollo embrionario relacionado con la delación de la región cromosómica 22q11,23. Se presenta en cerca de 1:4,000 nacimientos. deriva de la ausencia congénita del timo; anomalías cardíacas y renales, defectos faciales, hipoparatiroidismo, defectos esqueléticos y retraso en el desarrollo. El trastorno afecta a ambos sexos.

Inmunodeficiencia ligada al x con hiperinmunoglobulinemia M: grupo heterogéneo de trastornos por inmunodeficiencia primaria; derivan de insuficiencias de la recombinación para cambio de clase de las Ig durante la maduración de las células B, lo que conduce a una insuficiencia de IgG, IgA e IgE, pero elevadas concentraciones de IgM38. Sólo se presentan en varones. Los niños suelen presentar infecciones sinusales y pulmonares recurrentes, que pueden avanzar hasta desembocar en bronquiectasias y neumonía.

Inmunodeficiencias combinadas de células T y células B

Ataxia-telangiectasia: trastorno autosómico recesivo raro que deriva de la mutación de un gen (ATM), localizado en la región cromosómica 11 q22 23. Caracterizado por la neurodegeneración en el cerebelo y la telangiectasia oculocutánea. Se asocia con deficiencias inmunitarias, entre otras, linfopenia, hipogammaglobulinemia y disfunción inmunitaria mediada por células, que tienen como consecuencia el desarrollo recurrente de infecciones sinusales y pulmonares. Las personas con ese trastorno presentan un mayor riesgo de experimenta cáncer y sensibilidad a la radiación.

Síndrome de Wiskott-Aldrich: trastorno grave y complejo ligado al cromosoma x; se caracteriza por trombocitopenia, inmunodeficiencia, infecciones recurrentes, eccema, y aumento en el riesgo de desarrollar trastornos autoinmunitarios y linfomas.

Trastornos del sistema del complemento

Edema angioneurótico hereditario: manifestación que pone en riesgo la vida y puede conducir a la obstrucción completa de la vía respiratoria y la muerte si no se interviene. El edema de las estructuras de la mucosa gastrointestinal se relaciona con náuseas intensas, vómito y diarrea.

Bibliografía

1. Savides C., Shaker M. (2010). More than just infections: an update on primary immune deficiencies. Current Opinion in Pediatrics 22, 647–654. 2. Turvey S. E., Bonilla F. A., Junker A. K. (2009). Primary immunodeficiency diseases: A practical guide for clinicians. Postgraduate Medical Journal 85, 660–666. 3. Boyle J. M., Buckley R. H. (2007). Population prevalence of diagnosed primary immunodeficiency diseases in the United States. Journal of Clinical Immunology 27, 497–502. 4. Joshi A. Y., Iyer V. N., Hagan J. B., et al. (2009). Incidence and temporal trends of primary immunodeficiency: A population-based

cohort study. Mayo Clinical Proceedings 84, 16–22. 5. Notarangelo L. D., Fischer A., Geha R. S., et al. (2009). Primary immunodeficiencies: 2009 update. The Journal of Allergy and Clinical Immunology 124, 1161–1178. 6. Bonilla F. A., Geha R. S. (2009). Common variable immunodeficiency. Pediatric Research 65, 13R–9R. 7. Adeli M. M., Buckley R. H. (2010). Why newborn screening for severe combined immunodeficiency is essential: A case report. Pediatrics 126, e465–e469. 8. 10 warning signs of primary immunodeficiency 2011. Accessed September 9, 2011, at http://www.info4pi.org/aboutPI/index.cfm?section=aboutPI&content=warningsigns&CFID=67212&CFTOKEN=37730718. 9. Reda S. M., Afifi H. M., Amine M. M. (2009). Primary immunodeficiency diseases in Egyptian children: A single-center study. Journal of Clinical Immunology 29, 343–351. 10. Vassallo G., Newton R W ., Chieng S. E., et al. (2007). Clinical variability and characteristic autoantibody profile in primary C1q complement deficiency. Rheumatology (Oxford) 46, 1612–1614. 11. Keles S., Artac H., Kara R., et al. (2010). Transient hypogammaglobulinemia and unclassified hypogammaglobulinemia: 'Similarities and differences'. Pediatric Allergy and Immunology 21, 843–851. 12. Horwitz A., Kung S. J., McGeady S. J. (2010). Infants with low immunoglobulin levels: Isolated low IgA level vs other immunoglobulin abnormalities. Annals of Allergy, Asthma and Immunology 105, 295–298. 13. Duse M., Iacobini M., Leonardi L., et al. (2010). Transient hypogammaglobulinemia of infancy: Intravenous immunoglobulin as first line therapy. International Journal of Immunopathology and Pharmacology 3, 349–353. 14. Houlihan D. D., Storan E. R., Lee J. M. (2009). Sustained virologic response following HCV eradication in two brothers with Xlinked agammaglobulinaemia. World Journal of Gastroenterology 15, 3944–3946. 15. Winkelstein J. A., Conley M. E, James C., et al. (2008). Adults with X-linked agammaglobulinemia: Impact of disease on daily lives, quality of life, educational and socioeconomic status, knowledge of inheritance, and reproductive attitudes. Medicine 87, 253–258. 16. Kerns H. M., Ryu B. Y., Stirling B. V., et al. (2010). B cell-specific lentiviral gene therapy leads to sustained B-cell functional recovery in a murine model of X-linked agammaglobulinemia. Blood 115, 2146–2155. 17. Teimourian S., Nasseri S., Pouladi N., et al. (2008). Genotype-phenotype correlation in Bruton's tyrosine kinase deficiency. Journal of Pediatric Hematology/Oncology: official journal of the American Society of Pediatric Hematology/Oncology 30, 679–683. 18. Jacobs Z. D., Guajardo J. R., Anderson K. M. (2008). XLA-associated neutropenia treatment: A case report and review of the literature. Journal of Pediatric Hematology/Oncology: Official Journal of the American Society of Pediatric Hematology/Oncology 30, 631–634. 19. Salzer U., Maul-Pavicic A., Cunningham-Rundles C., et al. (2004). ICOS deficiency in patients with common variable immunodeficiency. Clinical Immunology 113, 234–240. 20. van Zelm M. C., Reisli I., van der Burg M., et al. (2006). An antibody-deficiency syndrome due to mutations in the CD19 gene. The New England Journal of Medicine 354, 1901–1912. 21. Bacchelli C., Buckridge S., Thrasher A. J., et al. (2007). Translational mini-review series on immunodeficiency: Molecular defects in common variable immunodeficiency. Clinical and Experimental Immunology 149, 401–409. 22. Giovannetti A., Pierdominici M., Mazzetta F., et al. (2007). Unravelling the complexity of T cell abnormalities in common variable immunodeficiency. Journal of Immunology 178, 3932. 23. Chapel H., Lucas M., Lee M., et al. (2008). Common variable immunodeficiency disorders: Division into distinct clinical phenotypes. Blood 112, 277–286. 24. Chua I., Quinti I., Grimbacher B. (2008). Lymphoma in common variable immunodeficiency: Interplay between immune dysregulation, infection and genetics. Current Opinion in Hematology 15, 368–374. 25. Saghafi S., Pourpak Z., Aghamohammadi A., et al. (2008). Selective immunoglobulin A deficiency in Iranian blood donors: Prevalence, laboratory and clinical findings. Iranian Journal of Allergy, Asthma, and Immunology 7, 157–162. 26. Bukowska-Strakova K., Kowalczyk D., Baran J., et al. (2009). The B-cell

compartment in the peripheral blood of children with different types of primary humoral immunodeficiency. Pediatric Research 66, 28–34. 27. Vorechovsky I., Zetterquist H., Paganelli R., et al. (1995). Family and linkage study of selective IgA deficiency and common variable immunodeficiency. Clinical Immunology and Immunopathology 77, 185–192. 28. Chowdary P., Nair D., Davies N., et al. (2010). Anaphylactic reaction with prothrombin complex concentrate in a patient with IgA deficiency and anti-IgA antibodies. Blood Coagulation and Fibrinolysis 21, 764–765. 29. Agarwal S., Cunningham-Rundles C. (2007). Assessment and clinical interpretation of reduced IgG values. Annals of Allergy, Asthma, and Immunology 99, 281–283. 30. Amariglio N., Lev A., Simon A., et al. (2010). Molecular assessment of thymus capabilities in the evaluation of T-cell immunodeficiency. Pediatric Research 67, 211–216. 31. Buckley R. H. (2004). Molecular defects in human severe combined immunodeficiency and approaches to immune reconstitution. Annual Review of Immunology 22, 625–655. 32. Fischer A., Hacein-Bey-Abina S., Cavazzana-Calvo M. (2011). Gene therapy for primary adaptive immune deficiencies. The

Journal of Allergy and Clinical Immunology 127, 1356–1359. 33. Balci Y. I., Turul T., Daar G., et al. (2008). Hematopoietic stem cell transplantation from a donor with Klinefelter syndrome for Wiskott-Aldrich syndrome. Pediatric Transplantation 12, 597–599. 34. McDonald-McGinn D. M., Sullivan K. E. (2011). Chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). Medicine (Baltimore) 90, 1–18. 35. Shaikh T. H., O'Connor R. J., Pierpont M. E., et al. (2007). Low copy repeats mediate distal chromosome 22q11.2 deletions: Sequence analysis predicts breakpoint mechanisms. Genome Research 17, 482–491. 36. Digilio M. C., McDonald-McGinn D. M., Heike C., et al. (2009). Three patients with oculo-auriculo-vertebral spectrum and microdeletion 22q11.2. American Journal of Medical Genetics 149A, 2860–2864. 37. McDonald-McGinn D. M., Kirschner R., Goldmuntz E., et al. (1999). The Philadelphia story: The 22q11.2 deletion: report on 250 patients. Genetic Counseling 10, 11–24. 38. Davies E. G., Thrasher A. J. (2010). Update on the hyper immunoglobulin M syndromes. British Journal of Haematology 149, 167–180. 39. Longo N. S., Lugar P. L., Yavuz S., et al. (2009). Analysis of somatic hypermutation in X-linked hyper-IgM syndrome shows specific deficiencies in mutational targeting. Blood 113, 3706–3715. 40. Winkelstein J. A., Marino M. C., Ochs H., et al. (2003). The X-linked hyper-IgM syndrome: Clinical and immunologic features of 79 patients. Medicine 82, 373–384. 41. Scott-Algara D., Balabanian K., Chakrabarti L. A., et al. (2010). Idiopathic CD4+ T-cell lymphocytopenia is associated with impaired membrane expression of the chemokine receptor CXCR4. Blood 115, 3708–3717. 42. Lipstein E. A., Vorono S., Browning M. F., et al. (2010). Systematic evidence review of newborn screening and treatment of severe combined immunodeficiency. Pediatrics 125, e1226–e1235. 43. Myers L. A., Patel D. D., Puck J. M., et al. (2002). Hematopoietic stem cell transplantation for severe combined immunodeficiency in the neonatal period leads to superior thymic output and improved survival. Blood 99, 872–878. 44. Kane L., Gennery A. R., Crooks B. N., et al. (2001). Neonatal bone marrow transplantation for severe combined immunodeficiency. Archives of Disease in Childhood. Fetal and Neonatal Edition 85, F110–F113. 45. Chan B., Wara D., Bastian J., et al. (2005). Long-term efficacy of enzyme replacement therapy for adenosine deaminase (ADA)deficient severe combined immunodeficiency (SCID). Clinical Immunology 117, 133–143. 46. Schmidt M., Hacein-Bey-Abina S., Wissler M., et al. (2005). Clonal evidence for the transduction of CD34+ cells with lymphomyeloid differentiation potential and self-renewal capacity in the SCID-X1 gene therapy trial. Blood 105, 2699–2706. 47. Hacein-Bey-Abina S., Garrigue A., Wang G. P., et al. (2008). Insertional oncogenesis in 4 patients after retrovirus-mediated gene therapy of SCID-X1. Journal of Clinical Investigation 118, 3132–3142. 48. Roberts J. L., Lengi A., Brown S. M., et al. (2004). Janus kinase 3 (JAK3) deficiency: Clinical, immunologic, and molecular analyses of 10 patients and outcomes of stem cell transplantation. Blood 103, 2009–2018. 49. Puel A., Ziegler S. F., Buckley R. H., et al. Defective IL7R expression in T (−) B (+) NK (+) severe combined immunodeficiency. Nature Genetics 20, 394–397. 50. Amorosi S., Russo I., Amodio G., et al. (2009). The cellular amount of the common gamma-chain influences spontaneous or induced cell proliferation. Journal of Immunology 182, 3304–3309. 51. Sauer A. V., Aiuti A. (2009). New insights into the pathogenesis of adenosine deaminase-severe combined immunodeficiency and progress in gene therapy. Current Opinion in Allergy and Clinical Immunology 9, 496–502. 52. Cassani B., Mirolo M.,

Cattaneo F., et al. (2008). Altered intracellular and extracellular signaling leads to impaired T-cell functions in ADA-SCID patients. Blood 111, 4209–4219. 53. Schwarz K., Gauss G. H., Ludwig L., et al. (1996). RAG mutations in human B cell-negative SCID. Science 274, 97–99. 54. Verhagen M. M., Abdo W . F., Willemsen M. A., et al. (2009). Clinical spectrum of ataxia-telangiectasia in adulthood. Neurology 73, 430–437. 55. Heath J., Goldman F. D. (2010). Idiopathic thrombocytopenic purpura in a boy with ataxia telangiectasia on immunoglobulin replacement therapy. Journal of Pediatric Hematology/Oncology: Official Journal of the American Society of Pediatric Hematology/Oncology 32, e25–e27. 56. Pietrucha B. M., Heropolitanska-Pliszka E., Wakulinska A., et al. (2010). Ataxia-telangiectasia with hyper-IgM and Wilms tumor: Fatal reaction to irradiation. Journal of Pediatric Hematology/Oncology: Official Journal of the American Society of Pediatric Hematology/Oncology 32, e28–e30. 57. Bosticardo M., Marangoni F., Aiuti A., et al. (2009). Recent advances in understanding the pathophysiology of Wiskott-Aldrich syndrome. Blood 113, 6288–6295. 58. Boztug K., Schmidt M., Schwarzer A., et al. (2010). Stem-cell gene therapy for the Wiskott-Aldrich syndrome. The New England Journal of Medicine 363, 1918–1927. 59. Ochs H. D., Thrasher A. J. (2006). The Wiskott-Aldrich syndrome. The Journal of Allergy and Clinical Immunology 117, 725– 738; quiz 39.