Instituto de Seguridad Social

GERENCIA

PRESTACIONES DE SALUD CRENADECER

UNIDAD DE

ANTENCION AMBULATORIA E INTERNACION

PAUTAS DE DIAGNÓSTICO, TRATAMIENTO Y SEGUIMIENTO

PARA

ERRORES INNATOS DEL METABOLISMO

Se adjuntan a continuación las principales citas bibliográficas de los protocolos existentes a la fecha para algunas de éstas enfermedades. Muchas de ellas no tienen protocolo específico y se utilizan las recomendaciones internacionales de grupos de expertos a nivel mundial.

Trastornos de Glicosilación de Proteínas

- Altassan R., Péanne R., Jaeken J., Barone R., Bidet M., et al. International clinical guidelines for the management of phosphomannomutase 2-congenital disorders of glycosylation: Diagnosis, treatment and follow up. J Inherit Metab Dis. 2019; 42: 5–28. DOI 10.1002/jimd.12024

Ciclo de Urea

- Häberle J., Boddaert N., Burlina A., Chakrapani A., Dixon M., Huemer M.,et al. Suggested guidelines for the diagnosis and management of urea cycle disorders. Orphanet Journal of Rare Diseases, 2012, 7:32.
  Disponible en: http://www.ojrd.com/content/7/1/32

Enfermedad de Fabry

- Concolino D., Degennarob E., Parini R. Delphi consensus on the current clinical and therapeutic knowledge on Anderson–Fabry disease. European Journal of Internal Medicine. 2014; 25: 751-756 DOI: 10.1016/j.ejim.2014.07.009

Aciduria Glutárica tipo I

- Boy N., Mühlhausen CH., Maier E., Heringer J.,et al. Proposed recommendations for diagnosing and managing individuals with glutaric aciduria type I: second revision. J Inherit Metab Dis, 2017; 40:75–101 DOI 10.1007/s10545-016-9999-9

Glucogenosis tipo I

- Kishnani P., Austin S., Abdenur J., Arn P., Bali D.. Diagnosis and management of glycogen storage disease type I: a practice guideline of the American College of Medical Genetics and Genomics. ACMG Standards and Guidelines, Genetics in Medicine, 2014; 16(11) DOI:10.1038/gim.2014.128

Niemann Pick tipo C

- Patterson M., Hendriksz Ch., Walterfang M., Sedel F., Vanier M., Wijburg F. on behalf of the NP-C Guidelines Working Group. Recommendations for the diagnosis and management of Niemann–Pick disease type C: An update. Molecular Genetics and Metabolism. 2012; 106: 330-3444[CZ1]

Tirosinemia tipo I

- Laet C., Dionisi-Vici C., Leonard J., McKiernan P., Mitchell G., et al.Recommendations for the management of tyrosinaemia type . Orphanet Journal of Rare Diseases 2013, 8:8. Disponible en: http://www.ojrd.com/content/8/1/8
- Chinsky J., Singh R., Ficicioglu C.,Karnebeek C., et al. Diagnosis and treatment of tyrosinemia type I: a US and Canadian consensus group review and recommendations. Genetics in Medicine, 2017; 19(12). DOI: 10.1038/gim.2017.101

Cobalamina C

- Huemer M., Diodato D., Schwahn., Schiff M., Bandeira A., et al. Guidelines for diagnosis and management of the cobalamin-related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency. J Inherit Metab Dis. 2017; 40:21–48 DOI 10.1007/s10545-016-9991-4

Acidemia metilmalónica y propiónica

- Baumgartner M., Hörster F., Dionisi-Vici C., Haliloglu G., Karall D., et al.Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemi Orphanet Journal of Rare Diseases 2014;9:130. Disponible en: http://www.ojrd.com/content/9/1/130

Galactosemia

- Welling L., Bernstein L.E., Berry G., Burlina A et al. International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up, J Inherit Metab Dis. 2017; 40:171–176 doi 10.1007/s10545-016-9990-5

3 Metil Crotonil CoA Carboxilasa deficiencia

- Arnold G., Koeberl D., Matern D-., Barshop B., Braverman N., et al- Delphi-based consensus clinical practice protocol for the diagnosis and management of 3-methylcrotonyl CoA carboxylase deficiency. Molecular Genetics and Metabolism. 2008: 93:363–370, DOI: 10.1016/j.ymgme.2007.11.002

Enfermedad mitocondrial

- Parikh S., Goldstein A., Koenig M, Scaglia F., Enns G., et al. Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genet Med. 2015; 17(9): 689–701. DOI:10.1038/gim.2014.177.

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- Pascual-Pascual SI, Nascimento A, Fernández-Llamazares CM, Medrano-López C et al. Clinical guidelines for infantile-onset Pompe disease. Rev Neurol. 2016; 63(6):269-79.
  Disponible en www.neurologia.com/articulo/2016232