Curriculum Vitae

Personal information

Francois Eyskens

Work experience

- AZ Middelheim- Children's Campus

1984 Assistant Training - Metabolic Diseases - General Pediatrics

- UZA - Pediatrics

1991 Consultant Metabolic Diseases (renewed on 01/01/2000)

- Provincial Centre for Detection of Metabolic Disorders - PCMA

1991 Lab Chief physician

1999 General Director

- General Children's Hospital Antwerp - AKA

1993 Consultant Paediatric

- Coordinating Committee and accreditation agency Beltest

1995 Representative of the Flemish Minister-President

- AZ Middelheim - Queen Paola Children's Hospital

1997 Fellow pediatrics

- Center for Inherited Metabolic Disorders

1998 Medical Coordinator

- PCMA

2003 Accreditation of the laboratory by PCMA Beltest / Belac.

Continued accreditation since then.

- ESN

2003 Representative of Flanders in the Board of ESN

(Inherited Metabolic Society of the Dutch language)

2007 Organizer of the Congress ESN Antwerp

- PCMA

2008 General Director of the Provincial Laboratory Centre

Detection of Metabolic Disorders in Antwerp

- UZA

July 2009 Pediatrics Clinic Head 6:10 appointment

- ZNA Queen Paola Children's Hospital

January 2010 Honorary Consultant Metabolic Diseases

- Medical Colleges to Be Expert in Drug NIHDI

Zavesca, Fabrazyme, Replagal, Aldurazyme, Myozyme, Elaprase.

- Belgian Guidelines for the Treatment of Gaucher Disease, 2005

  co-author

- Lysomed website, FOS, Myozyme database BOKS vzw

Medical Adviser

- Fabry International Study Group

Medical Expert

- King Baudouin Foundation

Member of the steering committee Rare Diseases

- National and international associations of Pediatrics, Metabolic Screening and

Member of several national and international associations of Pediatrics, and Metabolic Screening.

- Clinical Neurology and Neuro Surgery, European Journal of Pediatrics, Journal of Gastroenterology

Medical Review

- **The European Reference Network for Hereditary Metabolic Diseases (MetabERN)**

Member since April 2016

- **Metabolics.be (belgian scientific association of health providers with the objective of promotion of diagnosis, study and treatment of inborn errors of metabolism (IEM) of the children and the adults)**

President since 2019

- **Ethical Committee University Hospital Antwerp (UZA)**

Member since 2022

Education and training

UIA Doctor of medicine, 1984, surgery and obstetrics. Great distinction

UIA 04/12/1997: Ph.D. in medical sciences.

Thesis: "Inborn errors of metabolism in children. Epidemiological, clinical and biochemical aspects "

UIA 04/12/1997: Holder of the certificate Doctoral Studies (PhD).

Other

August 1989: Recognized specialist in pediatrics

Additional information

Publications

Publications (peer-reviewed journals):

1. Hendrickx J, Van Osta P, **Eyskens F**, et al;

Prenatal exclusion or Medium-Chain Acyl-CoA Dehydrogenase (MCAD) deficiency by direct detection of the white mutation PCR.

Diagnosis 1992 Jan; 12(1):74-76

2. **Eyskens F**, Van Doorn J, Mariën P;

Neurologic sequelae in transient nonketotic hyperglycinemia of the neonate.

J. Pediatr. 1992 Oct; 121(4):620-621

3. Van Acker K, **Eyskens F**, Verkerk R, Scharpé S;

Urinary excretion of purine and pyrimidine metabolites in the neonate.

Pediatric. Res. 1993 Dec, 34(6):762-766

4. *Workshop report: "Neonatal Screening for Duchenne Dystrophy" Consensus Recommendations of the 14 th workshop sponsored by the European Neuromuscular Centre*.

Neuromuscular Disorders 1993; Vol.3No.3, p.231-239, Co-author.

5. *Screening of Urinary purine and pyrimidine metabolites.* 4 th Symposium ESSPPMM. Nijmegen 1993. Abstract in Pharmacy World and Science suppl.F. 1993; Vol.15 No.4.

6. *Screening for Duchenne Dystrophy*,

Screening 1994; Vol.3No.3 :161-164, Co-author

7. **Eyskens F**, Ceuterick C, Martin JJ, Janssens G, Jaeken J;

Carbohydrate-deficient glycoprotein syndrome with previously unreported features.

Acta Paediatr. 1994 Aug; 83(8):892-896

8. Abstract in Enzyme and Protein. *"Central diabetes insipidus, adult respiratory distress syndrome and neutrophil dysfunction in a patient with combined respiratory chain defect*." 28 th Meeting of the European Metabolic Group. Utrecht 1995

9. **Eyskens F.**, Spaapen L.,

Hyperhomocysteinemia and Premature Vascular Disease.

Acta Neuro Logic Belgica 1996; 38

10. Van Acker K, **Eyskens F**, Espeel M, Wanders R, Dekker C, Kerckaert I, Roels F;

Hyperoxaluria with hyperglycoluria not due to alanine: glyoxylate aminotransferase defect: a novel type of primary hyperoxaluria.

Kidney International 1996 Nov, 50(5):1747-1752.

11. Pié J, Casals N, Casale C, C Bueas, Mascaró L, Barcelo A, Rolland MO, Zabot T, Haro D, **Eyskens F**, Divry P, Hegardt F

A nonsense mutation in the 3-hydroxy-3methylglutaryl-CoA (HMG-CoA) lyase gene produces exon skipping in two patients or different origin with HMG-CoA lyase deficiency.

Biochemical Journal 1997 Apr 15, 323 (2) :329-335

12. *Rickets: from nutritional problems to metabolic disease.*.

Opening Symposium Queen Paola Children's Hospital. Percentile, in December 1998, 3 (5) :146-151

13. Gibson KM, Sweetman L, Kozich V Pijackova A Tscharre A, **Eyskens F**, et al.

Unusual Enzyme Findings in Five Patients with metabolic profiles suggestive of succinic semialdehyde dehydrogenase deficiency (4-hydroxybutyric aciduria).

Inherit Metabolic Disorders 1998 jun, 21 (3) :255-61

14. Espeel M, **Eyskens F**, Wanders RJA, Lissens W, Roels F.

Peroxisome Mosaics in the Liver: the 7 th patient.

Cell Biology International 1998, 22 (1):64-65

15. **Eyskens F**, B Colfs.

The experience of the IRT screening or Cystic Fibrosis in the Province of Antwerp .Book: The International Experience of Screening for Cystic Fibrosis.

Proceedings of the Symposium on Screening for Cystic Fibrosis, 1998 CAEN

16. **Eyskens F**.

Screening for inborn errors of metabolism: the experience in the province of Antwerp. Summary doctoral thesis. Journal Pediatrics, May 1998

17. C.Mebis J Slabbynck H, Verhelst J, **Eyskens F**, Verrips A, Mahler C.

Cataract and xanthomen,

Journal of Medicine 1999, Vol 55 (11: 809-813)

18. Tanyalçin T, **Eyskens F**, M Lefevere, Philip E.

A marked difference between two Populations under mass screening of neonatal TSH and biotinidase activity. Accreditation and Quality Assurance 2002, 7 (11) :498-506

19. **Eyskens F**, Lefevere M.

Disturbances of valine metabolism in patient with peroxisomal disorders BioGenesis. Advances in Experimental Medicine and Biology 2003, 544:73-74

20. Roels F, Saudubray JM, Giros M, Handel H, Eyskens F, et al

Peroxisome mosaics,

Adv Exp Med Biol. 2003; 544:97-106

21. Tanyalçin T, **Eyskens F**, Phillips E, Lefevere M.

The use of split-sample design for performance evaluation or screening kits.

Accreditation and Quality Assurance 2004, 9 (3) :164-167

22. De Meirleir L, Seneca S, Lissens W, De Clercq I, **Eyskens F**, Gerlos E, et al,

Respiratory chain complex V deficiency due to a mutation in the assembly gene ATP12. Journal of Medical Genetics 2004, 41 (2) :120-124

23. Gootjes J, Elpeleg O, **Eyskens F**, Mandel H, Mitanchez D, et al,

Novel Mutations in the PEX2 gene of four unrelated Patients with a peroxisome biogenesis disorder.

Pediatric Research 2004 Mar, 55 (3) 431-436. Epub 2003 Nov 19

24. Jaeken J, Martens K, Francois I, **Eyskens F,** Lecointre C, Derua R, Meulemans S, et al,

Deletion of PREPL, a gene encoding a putative serine oligopeptidase, in patients with hypotonia-cystinuria syndrome.

Am J Hum Genet 2006 Jan; 78 (1): 38-51. Epub 2005 Nov 2003

25. Rooms L, Reyniers E, Wuyts W, Storm K, Van Luijk R, Scheers S, **Eyskens F**, et al,

Multiplex ligation-dependent probe amplification to detect subtelomeric rearrangements in routine diagnostics.

Clin Genet. 2006 Jan; 69(1): 58-64

26. Simons A, **Eyskens F**, De Groot A, Van Diest E, Deboutte D, Vermeiren R,

Cognitive Functioning of psychiatric disorders in children with a metabolic disease. European Child and Adolescent Psychiatry 2006 jun, 15 (4) :207-213. Epub 2006 Mar 10

27. Noelmans L, Jacquemyn Y, De Naeyer S, **Eyskens F**,

Pregnancy and galactosaemia

J Obstet Gynaecol. 2006 Nov; 26(8): 812-4

28. Van Bever Y, Balemans W, Duval EL, Jespers A, **Eyskens F,** Van Hul W, Courtens W

Exclusion of OGDH and BMP4 as candidate genes in 2 siblings with autosomal recessive DOOR syndrome.

Am J Med Genet A. 2007 Apr 1; 143A(7): 763-7

29. Brouns R, **Eyskens F**, De Deyn P.

Middelheim Fabry Study (MiFaS): A retrospective Belgian study of the prevalence of Fabry disease in young Patients with cryptogenic stroke.

Clinical Neurology and Neurosurgery 2007 Jul; 109(6):479-84. Epub 2007 May 16.

30. . Boudewijns, A., Declau, F., Smets, K., Ursi, D., **Eyskens, F**., et al

Cytomegalovirus DNA detection in Guthrie cards: Role in the diagnostic work-up of childhood hearing loss,

Otology and Neurotology 2009 Oct, 30 (7), 943-949.

31. Roels, F., Verloo, P., **Eyskens, F**., Francois, B., Seneca, S., De Paepe, B., et al.

Mitochandrial Mosaics in the liver of 3 infants with mtDNA defects.

BMC Clinical Pathology 2009 Jun 5; 9:4.

32. Ebberink, MS, Mooyer, PAW, Koster, J., Dekker, CJM, **Eyskens, F**, et al,

Genotype-phenotype correlation in Pex5-deficient peroxisome bioGenesis defective cell lines.

Human Mutation 2009 Jan, 30 (1), 93-98.

33. **F. Eyskens**,

Rare inborn errors of metabolism in adults: The lysosomal storage disorders.

Acta Clinica Belgica 2009 Nov-dec; 64; (6), 534-539.

34. Brouns, R., **Eyskens, F**., De Boeck, K., Ceuterick-de Groote, C., et al,

Fabry disease in a patient with Turner syndrome.

J Inherited Metabolic Disease 2009 Dec, 32 Suppl 1:S45-48

35. Brouns, R., Thijs, V., **Eyskens, F**., Van Den Broeck, M., Belachew, S., et al

Belgian Fabry study: Prevalence of Fabry disease in a cohort of 1000 young Patients with cerebrovascular disease,

Stroke 2010 may, 41 (5), 863-868.

36. Kuilenburg, ABP, Dobritzsch, D., Meijer, J., **Eyskens, F**. et al;

Dihydropyrimidinase deficiency: Phenotype, genotype and structural consequences in 17 patients,
Press.Biochimica et Biophysica Acta - Molecular Basis of Disease 2010 jul-aug; 1802(7-8):639-648.

37. Mehta A, Beck M, **Eyskens F** et al.

Fabry Disease: a review of current management strategies.

QJM 2010 sep; 103 (9): 641-59

38. Van den Bulcke T, Vanden Broucke P, Van Hoof V, **Eyskens F**, et al

Data mining methods for classification of Medium-Chain Acyl-CoA dehydrogenase deficiency (MCADD) using non-derivatized tandem MS neonatal screening data.

J Biomed Inform. 2011 Apr;44(2):319-25.

39. De Brabander I, Yperzeele L, Ceuterick-De Groote C, **Eyskens F**, et al

Phenotypical characterization of α-galactosidase A gene mutations identified in a large Fabry disease screening program in stroke in the young.

Clin Neurol Neurosurg. 2013 Jul;115(7):1088-93. doi: 10.1016/j.clineuro.2012.11.003. Epub 2012 Dec 4.

40. Vandevijvere S, Coucke W, Vanderpas J, **Eyskens F**,et al.

Neonatal thyroid-stimulating hormone concentrations in Belgium: a useful indicator for detecting mild iodine deficiency?

PLoS One. 2012;7(10):e47770. doi: 0.1371/journal.pone.0047770. Epub 2012 Oct 24.

41. Ajit Bolar N, Vanlander AV, Wilbrecht C, **Eyskens F**, et al

Mutation of the iron-sulfur cluster assembly gene IBA57 causes severe myopathy and encephalopathy.

Hum Mol Genet. 2013 Jul 1;22(13):2590-602. doi: 10.1093/hmg/ddt107. Epub 2013 Mar 5.

42. Warnock, D; Bichet, D; Holida, M; **Eyskens, F** et al

Phase2a study to investigate the effect of a single dose of migalastat HCL, a pharmacological chaperone, on agalsidase activity in subjects with Fabry disease. Molecular genetics and metabolism 2013, 108 (2): S96-S96, WOS:000314670500250

43. Adam S, Almeida MF, Carbasius Weber E, **Eyskens F**, et al

Dietary practices in pyridoxine non-responsive homocystinuria: a European survey.

Mol Genet Metab. 2013 Dec;110(4):454-9. doi: 10.1016/j.ymgme.2013.10.003. Epub 2013 Oct 10.

44. Adam S, Almeida MF, Assoun M, **Eyskens F**, et al.

Dietary management of urea cycle disorders: European practice.

Mol Genet Metab. 2013 Dec;110(4):439-45. doi: 10.1016/j.ymgme.2013.09.003. Epub 2013 Sep 12.

45. Luyckx E, **Eyskens F**, Simons A, Beckx K, Van West D, Dhar M;

Long-term follow-up on the effect of combined therapy of bile acids and statins in the treatment of cerebrotendinous xanthomatosis: a case report.

Clin Neurol Neurosurg. 2014 Mar;118:9-11. doi: 10.1016/j.clineuro.2013.12.008. Epub 2013 Dec 29.

46. Jacquemyn Y, Den Hartog M, **Eyskens F**.

Methylmalonic acidaemia in pregnancy.

BMJ Case Rep. 2014 Mar 31;2014. pii: bcr2014203723. doi: 10.1136/bcr-2014-203723

47. Warnock DG, Bichet DG, Holida M, **Eyskens F**, et al.

Oral Migalastat HCl Leads to Greater Systemic Exposure and Tissue Levels of Active α-Galactosidase A in Fabry Patients when Co-Administered with Infused Agalsidase.

PLoS One. 2015 Aug 7;10(8):e0134341. doi: 10.1371/journal.pone.0134341. eCollection 2015

48. De Bruyn A, Jacquemyn Y, Kinget K, **Eyskens F**.

Carnitine Deficiency and Pregnancy.

Case Rep Obstet Gynecol. 2015;2015:101468. doi: 10.1155/2015/101468. Epub 2015 May 28.

49. Gatheridge MA, Kwon JM, Mendell JM, **Eyskens F**, et al.

Identifying Non-Duchenne Muscular Dystrophy-Positive and False Negative Results in Prior Duchenne Muscular Dystrophy Newborn Screening Programs: A Review.

JAMA Neurol. 2016 Jan 1;73(1):111-6. doi: 10.1001/jamaneurol.2015.3537

50. Van Ginkel WG, Jahja R, Huijbregts SC, **Eyskens F**, et al

Neurocognitive outcome in tyrosinemia type 1 patients compared to healthy controls.

Orphanet J Rare Dis. 2016 Jun 29;11(1):87. doi: 10.1186/s13023-016-0472-5.

51. Hughes DA, Nicholls K, Shankar SP, **Eyskens F**, et al.

Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study.

J Med Genet. 2016 Nov 10. pii: jmedgenet-2016-104178. doi: 10.1136/jmedgenet-2016-104178.

52. Van Erven B[1], Welling L[2], van Calcar SC[3], Doulgeraki A[4], **Eyskens F**, et al.

Bone Health in Classic Galactosemia: Systematic Review and Meta-Analysis.

JIMD Rep. 2016 Dec 20.

53. Simons A, **Eyskens F**, Glazemakers I, van West D.

Can psychiatric childhood disorders be due to inborn errors of metabolism?

Eur Child Adolesc Psychiatry. 2017 Feb;26(2):143-154. doi: 10.1007/s00787-016-0908-4. Epub 2016 Sep 30.

54. Pinto A, Daly A, Evans S, **Eyskens F**, et al.

Dietary practices in isovaleric acidemia: A European survey.

Mol Genet Metab Rep. 2017 Feb 27;12:16-22. doi: 10.1016/j.ymgmr.2017.02.001. eCollection 2017.

55. Welling L, Bernstein LE, Berry GT, **Eyskens F**, Galactosemia Network (GalNet), et al.

International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up.

J Inherit Metab Dis. 2017 Mar;40(2):171-176. doi: 10.1007/s10545-016-9990-5. Epub 2016 Nov 17

56. Muntau AC, Burlina A, **Eyskens F**, et al.

Efficacy, safety and population pharmacokinetics of sapropterin in PKU patients <4 years: results from the SPARK open-label, multicentre, randomized phase IIIb trial.

Orphanet J Rare Dis. 2017 Mar 9;12(1):47. doi: 10.1186/s13023-017-0600-x.

57. Vanacoleyen T, Van Den Sande A, Vanderstraeten W, **Eyskens F**, et al.

Applicability and desirability of screening for disease of Duchenne and disease of Pompe.

Tijdschrift voor Geneeskunde, Sep 2017. 73(18):1093-1104.

58. Daly A, Pinto A, Evans S, **Eyskens F**, et al

Dietary practices in propionic acidemia: A European survey.

Mol Genet Metab Rep. 2017 Oct 3;13:83-89. doi: 10.1016/j.ymgmr.2017.09.002. eCollection 2017 Dec.

59. **Eyskens F**, Devos S.

Newborn Screening for Lysosomal Storage Disorders in Belgium: The importance of Sex- and Age-Dependent Reference Ranges.

Journal of Inborn Errors of metabolism and screening 2017. Volume 5: 1-8.

60. Bonham J, Carling R, Lindner M, Franzson L, **Eyskens F**, et al.

Raising awareness of false positive newborn screening results arising from pivalate-containing creams and antibiotics in Europe when screening for Isovaleric Acidaemia.

Int. J. Neonatal Screen. 2018, 4, 8; doi: 10.3390

61. **Eyskens F**.

Newborn screening for lysosomal diseases: the need for sex and gestational age dependent reference ranges.

Molecular Genetics an Metabolism 123(2): S43-S44. Feb 2018.

62. Hauth L, Kerstens J, Yperzeele L, **Eyskens F**, Parizel PM, Willekens B *Galactosidase Alpha p.A143T Variant Fabry Disease May Result in a Phenotype With Multifocal Microvascular Cerebral Involvement at a Young Age.*

Front Neurol. 2018 May 16;9:336. doi: 10.3389/fneur.2018.00336. eCollection 2018.

63. Simons A, **Eyskens F**, Raets E, et al.

Girl with Tyrosinemia Type 1 and Executive dysfunctions treated with Methylphenidate: a case report.

Journal of Inborn Errors of Metabolism and Screening. 2018, Vol 6:1-3.

64. Posset R, Garbade SF, Boy N, et al; Additional individual contributors of the UCDC and the E-IMD consortium.

Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders-A successful strategy for clinical research of rare diseases.

J Inherit Metab Dis. 2019 Jan;42(1):93-106. doi: 10.1002/jimd.12031.

65. Molema F, Gleich F, Burgard P, et al; Additional individual contributors from E-IMD.

Evaluation of dietary treatment and amino acid supplementation in organic acidurias and urea-cycle disorders: On the basis of information from a European multicenter registry.

J Inherit Metab Dis. 2019 Feb 8. doi: 10.1002/jimd.12066.

66. Rubio-Gozalbo ME, Haskovic M, Bosch AM, **Eyskens F**, et al.

The natural history of classic galactosemia: lessons from the GalNet registry.

Orphanet J Rare Dis. 2019 Apr 27;14(1):86. doi: 10.1186/s13023-019-1047-z.

67. Molema F, Gleich F, Burgard P, et al; Additional individual contributors from E-IMD.

Decreased plasma l-arginine levels in organic acidurias (MMA and PA) and decreased plasma branched-chain amino acid levels in urea cycle disorders as a potential cause of growth retardation: Options for treatment.

Mol Genet Metab. 2019 Apr;126(4):397-405. doi: 10.1016/j.ymgme.2019.02.003. Epub 2019 Feb 25.

68. Heard JM, Bellettato C, van Lingen C, Scarpa M; MetabERN collaboration group.

Research activity and capability in the European reference network MetabERN.

Orphanet J Rare Dis. 2019 May 29;14(1):119. doi: 10.1186/s13023-019-1091-8.

69. Nestele J, van den Hoven C, Van Craenenbroeck EM, **Eyskens F**, Paelinck BP, Dendooven A, Haine SE.

Recurrent acute coronary syndrome, polymorphic premature ventricular complexes and a son with a (mis)diagnosis of multiple sclerosis.

Acta Cardiol. 2019 May 16:1-2. doi: 10.1080/00015385.2019.1616137.

70. Posset R, Gropman AL, Nagamani SCS, et al; Urea Cycle Disorders Consortium and the European Registry and Network for Intoxication Type Metabolic Diseases Consortia Study Group.

Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders.

Ann Neurol. 2019 Jul;86(1):116-128. doi: 10.1002/ana.25492. Epub 2019 May 13.

71. Smets F, Dobbelaere D, McKiernan P, **Eyskens F**, et al.

Phase I/II Trial of Liver-derived Mesenchymal Stem Cells in Pediatric Liver-based Metabolic Disorders: A Prospective, Open Label, Multicenter, Partially Randomized, Safety Study of One Cycle of Heterologous Human Adult Liver-derived Progenitor Cells (HepaStem) in Urea Cycle Disorders and Crigler-Najjar Syndrome Patients.

Transplantation. 2019 Sep;103(9):1903-1915. doi: 10.1097/TP.0000000000002605.

72.Van Vliet K, van Ginkel WG, Jahja R, Daly A, MacDonald A, Eyskens F, et al. *Emotional and behavioral problems, quality of life and metabolic control in NTBC-treated Tyrosinemia type 1 patients.*

Orphanet J Rare Dis. 2019 Dec 4;14(1):285. doi: 10.1186/s13023-019-1259-2.

73. Molema F, Gleich F, Burgard P, van der Ploeg AT, Summar ML, Chapman KA, Barić I, Lund AM, Kölker S, Williams M; Additional individual contributors from E-IMD.

Evaluation of dietary treatment and amino acid supplementation in organic acidurias and urea-cycle disorders: On the basis of information from a European multicenter registry.

J Inherit Metab Dis. 2019 Nov;42(6):1162-1175. doi: 10.1002/jimd.12066.

74. Pinto A, Evans S, Daly A et al.

Dietary practices in methylmalonic acidemia. An European survey.

Journal of Pediatric Endocrinology and Metabolism. 2020; 33(1): 147-155.

75. Eyskens F.

The Belgian Hematology Project.

Molecular Genetics and Metabolism 2020; 129(2): S54-S55.

76. Libbrecht S, Eyskens F, Ceclercq S, Colpaert C.

Placental Findings in Lysosomal Storage Disease Diagnosis: A case report of Galactosialidosis.

Case Reports in Pathology 2020.

77. Posset R, Garbade SF, Gleich F, Gropman AL, de Lonlay P, Hoffmann GF, et al.

Urea Cycle Disorders Consortium (UCDC); European registry and network for Intoxication type Metabolic Diseases (E-IMD).

Long-term effects of medical management on growth and weight in individuals with urea cycle disorders.

Sci Rep. 2020 Jul 20;10(1):11948. doi: 10.1038/s41598-020-67496-3.

78. Nestele J, van den Hoven C, Van Craenenbroeck EM, Eyskens F, Paelinck BP, et al.

Recurrent acute coronary syndrome, polymorphic premature ventricular complexes and a son with a (mis)diagnosis of multiple sclerosis.

Acta Cardiol. 2020 Sep;75(5):467-468. doi: 10.1080/00015385.2019.1616137. Epub 2019 May 16.

79. Vandevelde NM, Vermeersch P, Devreese KMJ, Vincent MF, Gulbis B, Eyskens F et al.

Belgian rare diseases plan in clinical pathology: identification of key biochemical diagnostic tests and establishment of reference laboratories and financing conditions.

https://doi.org/10.21.203/rs3-rs35562/v1

80. Muntau AC, Burlina A, Eyskens F, Freisinger P, Leuzzi V, Sivri HS, Gramer G et al.

Long-term efficacy and safety of sapropterin in patients who initiated sapropterin at < 4 years of age with phenylketonuria: results of the 3-year extension of the SPARK open-label, multicentre, randomised phase IIIb trial.

Orphanet J Rare Dis. 2021 Aug 3;16(1):341. doi: 10.1186/s13023-021-01968-1.

81. Landen M, Eyskens F, Vanhoenacker F.

Vertebral Tongue-Like Deformity in Mucopolysaccharidosis VI.

J Belg Soc Radiol. 2021 Sep 27;105(1):54. doi: 10.5334/jbsr.2611. eCollection 2021.

82. Van Vliet K, Van Ginkel WG, Jahja R, Daly A, MacDonald A, Eyskens F, et al. *Neurocognitive outcome and mental health in children with tyrosinemia type 1 and phenylketonuria: A comparison between two genetic disorders affecting the same metabolic pathway.* J Inherit Metab Dis. 2022 Sep;45(5):952-962. doi: 10.1002/jimd.12528. Epub 2022 Jun 30.

83. Eyskens M, Bruyndonckx L, Van Kuilenburg ABP, Eyskens F. Severe dilated cardiomyopathy as an unusual clinical presentation in an infant with sialidosis type II. JIMD Rep. 2023 Jan 7;64(2):156-160
84. Metachromatic leukodystrophy: To screen or not to screen? Jonckheere AI, Kingma SDK, Eyskens F, Bordon V, Jansen AC. Eur J Paediatr Neurol. 2023 Sep;46:1-7
85. A new multiplex analysis of glucosylsphingosine and globotriaosylsphingosine in dried blood spots by tandem mass spectrometry. Van Baelen A, Roosens L, Devos S, Verhulst S, Eyskens F. Mol Genet Metab Rep. 2023 Aug 18;37
86. Unexplained splenomegaly as a diagnostic marker for a rare but severe disease with an innovative and highly effective new treatment option: A case report. Van Baelen A, Verhulst S, Eyskens F. Mol Genet Metab Rep. 2024 Sep 28;41
87. Left ventricular hypertrophy: do not forget Fabry disease. Diagnostic work-up and differential diagnosis. Paelinck BP, Bondue A, Robyns T, Eyskens F. Acta Cardiol. 2024 Aug;79(6):642-649
88. A phase III, open-label clinical trial evaluating pegunigalsidase alfa administered every 4 weeks in adults with Fabry disease previously treated with other enzyme replacement therapies. Holida M, Linhart A, Pisani A, Longo N, Eyskens F, et al. J Inherit Metab Dis. 2025 Jan;48(1):e12795
89. High-Resolution Mass Spectrometry Method for Targeted Screening and Monitoring of Fabry, Gaucher and ASM Using Dried Blood Spots and Capitainers: Impact of Sample Matrix on Measurements Results. Van Baelen A, Verhulst S, Eyskens F. Int J Mol Sci. 2025 Aug 7; 26(15): 7641

Projects

Coordinator of WP 6 Learning&Training of the European Reference Network MetabERN Consortium 2023-2027.

Coordinator/PI of the Pilot project Home Therapy Enzyme Replacement Therapy of Lysosomal Storage Diseases part of Transmural Care for Children affected by a Chronic Disease (Ministry of Health/FOD).

Memberships	MetabERN (see above) SSIEM ISNS FAGG

Other Relevant Information