



## Profolio of Researchers and Research Teams

### GI-1345 NEONATAL AND DEVELOPMENT MEDICINE

(Last update 03/04/2013)

**Code:** GI-1345  
**Department:** Pediatría  
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#### Research field

- Inborn Errors of Metabolism, newborn screening, diagnosis, treatment, metabolic
- Neonatal lung physiology, high frequency ventilation, surfactant, nitric oxid, biophysic monitoring
- Newborn Nutrition, Oligoelements, Trace Elements, bioavailability and speciation, "in vitro" digestion, Analytical Methods (with GI 1622)
- Epidemiology and treatment of Neonatal Infections
- Development, growth and global assessment, , neonatal sonography

#### Technology services

- Neonatal Screening,  
Tandem Mass Spectrometry,  
Analysis of Aminoacids, Carnitines and Trace Elements  
Physiology of Respiration

## Key words

Neonatology;  
Pediatrics;  
Growth;  
Neonatal Screening;  
Metabolism;  
Trace Elements;  
Tandem Mass Spectrometry;  
Atomic Absorption Spectrometry; Aminoacid analysis

## Researchers

<b>Name</b>	<b>Position</b>
Fraga Bermúdez, J.M.	Coordinator
Alonso Fernández, J.R.	Member
Couce Pico, M.L.	Member
Fernández Lorenzo, J.R.	Member
Martínez Soto, M.I.	Member
Varela Iglesias, J.J.	Member

### **External collaborators**

José Ángel Cocho de Juan	CHUS
Dolores Bóveda Fontán	CHUS
Daisy Emilia Castiñeiras Ramos	CHUS
Cristóbal Colon Mejeras	CHUS
Maria José Fernandez Seara	CHUS
MARTA GIL CALVO	CHUS
A. Javier Iglesias Rodríguez	CHUS
Alejandro Pérez Muñuzuri	CHUS
María Mayra Rebolledo Fernández	IDICHUS-REDEMETH
Ana Fernández Marmiesse	FGMG
Ana María Baña Souto	CHUS
Olalla Elena López Suárez	CHUS
Paula Sánchez Pintos	SERGAS

## Technological customers and partners

- Genzyme S.L.
- Sanofi Aventis, S.A.

## SCIENTIFIC PRODUCTION ( period: 2007 - 2011 )

### **Articles in scientific journals**

#### **Article:**

The tetrahydrobiopterin loading test in 36 patients with hyperphenylalaninaemia: evaluation of response and subsequent treatment

**Journal:** JOURNAL OF INHERITED METABOLIC DISEASE, ISSN: 0141-8955  
2007

#### **Article:**

Hypermethioninaemia due to methionine adenosyltransferase I/III (MAT I/III) deficiency: diagnosis in an expanded neonatal screening programme.

**Journal:** JOURNAL OF INHERITED METABOLIC DISEASE, ISSN: 0141-8955  
2008

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**Article:**

The contributions of Louis I Woolf to the treatment, early diagnosis and understanding of phenylketonuria

**Journal:** JOURNAL OF MEDICAL SCREENING, ISSN: 0969-1413  
2009

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**Article:**

Isolated growth hormone deficiency type II caused by a novel intronic mutation

**Journal:** HORMONE RESEARCH, ISSN: 0301-0163  
2009

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**Article:**

Serum levels of IGF1 are a useful predictor of retinopathy of prematurity.

**Journal:** ACTA PAEDIATRICA, ISSN: 0803-5253  
2010

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**Article:**

Hyperammonaemia. Treatment in the emergency and acute phase of a patient with citrullinaemia.

**Journal:** ANALES DE PEDIATRIA, ISSN: 1695-4033  
2010

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**Article:**

Hypermethioninemia in the preterm newborn. Predisposing factors.

**Journal:** ANALES DE PEDIATRIA, ISSN: 1695-4033  
2010

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**Article:**

Vertical sandwich-type continuous/evaporative TLC with fixed mobile phase volume for separating sugars of clinical relevance in paper-borne urine and blood samples in newborn screening.

**Journal:** JOURNAL OF CLINICAL LABORATORY ANALYSIS, ISSN: 0887-8013  
2010

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**Article:**

Neonatal screening for mucopolysaccharidoses by determination of glycosaminoglycans in the eluate of urine-impregnated paper: preliminary results of an improved DMB-based procedure.

**Journal:** JOURNAL OF CLINICAL LABORATORY ANALYSIS, ISSN: 0887-8013  
2010

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**Article:**

Newborn screening in Spain, with particular reference to Galicia: echoes of Louis I. Woolf.

**Journal:** MOLECULAR GENETICS AND METABOLISM, ISSN: 1096-7192  
2010

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**Article:**

Evolutionary Analyses of Entire Genomes Do Not Support the Association of mtDNA Mutations with Ras/MAPK Pathway Syndromes.

**Journal:** PLoS One, ISSN: 1932-6203  
2011

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**Article:**

The TSH threshold in neonatal screening for congenital hypothyroidism: a variable solution.

**Journal:** ARCHIVES OF DISEASE IN CHILDHOOD, ISSN: 0003-9888  
2011

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**Book:**

**Title:**

Fundamentos de Economía y Gestión de la Salud  
(978-84-9887-532-4) Publicación: 2010

**Editorial:** Servizo de publicacións e intercambio científico

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**Chapter:**

**Title:**

Oxigenoterapia y soporte ventilatorio

**Book:**

Tratado de Neumología Infantil. 2ª Edición

(978-84-8473-719-3) Publicación: 2008

**Editorial:** Ergon

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**Title:**

Enfermedad congénita del metabolismo: signos de sospecha y abordaje terapéutico inicial.

**Book:**

De guardia en Neonatología

(978-84-8473-626-4) Publicación: 2008

**Editorial:** Ergon

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**Doctoral theses:**

**Title:**

Desenvolvemento e aplicación de métodos de análise por espectrometría de masas en tándem para o estudo de metabolitos urinarios marcadores de erros conxénitos do metabolismo

**Date of dissertation:** 03/12/2007

**Director:** José María Fraga Bermúdez, Pilar Bermejo Barrera

**Author:** María Mayra Rebolledo Fernández

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**Title:**

Desarrollo de un método por espectrometría de masas en tándem para la determinación de acilcarnitinas y la detección neonatal de alteraciones del metabolismo de ácidos orgánicos y ácidos grasos

**Date of dissertation:** 16/10/2007

**Director:** José María Fraga Bermúdez, María Herminia Bollaín Rodríguez

**Author:** José Ángel Cocho de Juan

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Desenvolvemento e aplicación de métodos de análise por espectrometría de masas en tándem para o estudo de metabolitos urinarios marcadores de erros conxénitos do metabolismo

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**Date of dissertation:** 16/10/2007

**Director:** José María Fraga Bermúdez, María Herminia Bollaín Rodríguez

**Author:** José Ángel Cocho de Juan

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**Title:**

Retinopatía de la prematuridad: estudio prospectivo en recién nacidos de menos de 32 semanas y/o menos de 1500 gr sobre los factores etiopatogénicos. Utilidad clínica y diagnóstica de la medición doppler de la arteria central de la retina y de la determinación secuencial de los niveles del factor de crecimiento similar a la insulina tipo 1

**Date of dissertation:** 07/07/2009

**Director:** José María Fraga Bermúdez, José Ramón Fernández Lorenzo

**Author:** Alejandro Pérez Muñuzuri

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