

ÉTICA Y NUEVAS TECNOLOGÍAS EN CLÍNICA PEDIÁTRICA

Manuel Moya. UMH/ HUSJ

Presidente del Comité Ético de
la European Pediatric
Association :

- 41 Sociedades Nacionales de Europa
- >60000 pediatras

ETHIKOS, nacido de la costumbre para determinar lo apropiado o lo erróneo

La ética (John Dewey)

- Es un concepto y no un modo de ser o una condición humana universal.
- Está sujeta a cambios que siguen al conocimiento científico y que socialmente requieren tiempo (generaciones)

ÉTICA Y NUEVAS TECNOLOGÍAS

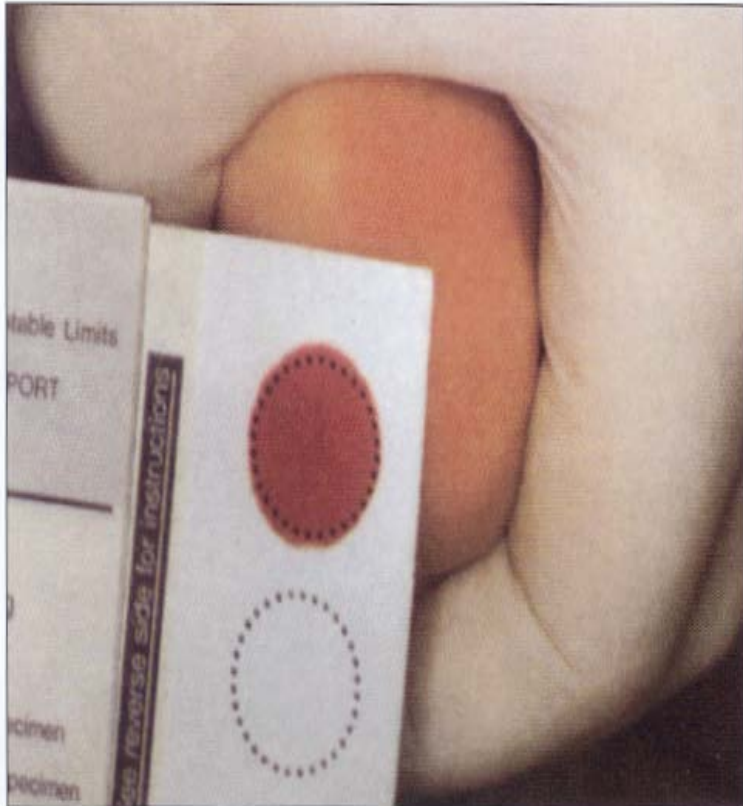
1 Screening neonatal universal

2 Variaciones del número de copias del DNA

3 Uso de tejidos humanos e investigación etiológica de enfermedades.

4 Tecnologías reproductivas

SCREENING NEONATAL UNIVERSAL



PKU
HIPOTIROIDISMO C

(Ac VIH)

7 Toque la gota GRANDE de sangre levemente con el papel de filtro. Deje que la sangre se absorba y que llene el círculo por completo con una SOLA aplicación a la gota GRANDE de sangre. (Para aumentar el flujo de sangre, puede aplicar presión MUY LEVE en forma intermitente al área que rodea el sitio de punción.) Aplique la sangre solamente a uno de los lados del papel de filtro.

SCREENING NEONATAL. ESPECTROMETRÍA DE MASAS EN TANDEM (MS/MS)

ACMG/ASHG statement

Table 1

Some disorders detectable by tandem mass spectrometry

Disorder	Diagnostic metabolite
Amino acidemias	
Phenylketonuria	Phenylalanine & tyrosine
Maple syrup urine disease	Leucine + isoleucine
Homocystinuria (CBS deficiency)	Methionine
Citrullinemia	Citrulline
Hepatorenal tyrosinemia	Methionine & tyrosine
Organic acidemias	
Propionic acidemia	C ₃ acylcarnitine
Methylmalonic acidemia(s)	C ₃ acylcarnitine
Isovaleric acidemia	Isovalerylcarnitine
Isolated 3-methylcrotonylglycinemia	3-Hydroxyisovalerylcarnitine
Glutaric acidemia (type I)	Glutarylacarnitine
Hydroxymethylglutaric acidemia	Hydroxymethylglutarylacarnitine
Fatty acid oxidation disorders	
SCAD deficiency	C _{4,6} acylcarnitines
MCAD deficiency	C _{8,10,12} acylcarnitines
VLCAD deficiency	C _{14,16,18,20} acylcarnitines
LCHAD and trifunctional protein deficiency	C _{14,16,18,20} acyl- and 3-hydroxy acylcarnitines
Glutaric acidemia type II	Glutarylacarnitine
CPT-II deficiency	C _{14,16,18,20,22} acylcarnitines

2000:

10 uL 2' >35 metabolitos

2011:

11 metabolitos + FQ+ Galac +...

Testing Children for Adult-Onset Genetic Diseases

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KEY WORDS

genetics, ethics, policy, screening, breast cancer, polyps

The Committee on Bioethics of the American Academy of Pediatrics (AAP) advises against genetic testing of children for conditions that have their onset in adulthood. They write, "Genetic testing for adult-onset conditions generally should be deferred until adulthood or until an adolescent interested in testing has developed mature decision-making capacities."¹ The Clinical Genetics Society in the United Kingdom expresses similar views, writing that "The predictive genetic testing of children...for an adult onset disorder should generally not be undertaken...until the 'children' request such tests for themselves, as autonomous adults."²

VARIACIONES DEL NÚMERO DE COPIAS (CNVs) DEL DNA DE UNA REGIÓN DEL GENOMA

Métodos: micromatrices cromosómicas y para SNPs,
hibridación genómica comparativa

CNVs: Patológicas son más grandes y generalmente 'de novo'

SNPs: Paternidad, duplicaciones, gene FTO...

Permiten descubrir aberraciones genotípicas (cariotipo normal)
que puedan correlacionarse con anomalías fenotípicas

TABLE 1 Clinically Significant Genes and Loci Implicated in the Development of Isolated Structural Birth Defects as Determined by CNV Identification

Structural Birth Defect	Disease Incidence at Birth, %	Cases With Putative Causal CNVs, %	Implicated Gene/Loci
Craniofacial defects			
Cleft lip and/or palate	0.1	N/A	<i>SUM01</i>
Sotos syndrome	0.007	15.0	<i>PLXDC2, NPAS3</i>
NMLFS	<0.0001	N/A	8q22.1 region
Single suture craniosynostosis	0.04–0.1	7.5	<i>RUNX2</i>
Respiratory defects			
CDH		10.3	15q21.1-q26.2 region
	0.02–0.05		<i>COUP-TFII</i>
Renal Defects			
Congenital renal and UT anomalies	0.2	10.0	<i>SOX17</i>
Cardiac Defects			
TOF	0.03	1.0	1q21.1 region

NA, not available.

Southard AE et al.

Role of Copy Numbers Variants in Structural Birth Defects.

Pediatrics 2012; 129: 755-62

VARIACIONES DEL NÚMERO DE COPIAS Y CUESTIONES SUSCITADAS

Precisión del μ array > metafase

Cuadro malformativo + CNV (grande)

CNV a los padres

-vo

de novo

+vo

Información genética

USO DE TEJIDOS HUMANOS PARA INVESTIGACIÓN ETIOLÓGICA DE ENFERMEDADES (Biobanking)

Fundamento: Provisión de especímenes a científicos que investigan enfermedades

- **Obtención de especímenes limitada en niños.**

- **Sólo biobancos académicos y con la infraestructura requerida**

Brisson AL et al. Pediatrics 2012; 129: 153-62

ESPECÍMENES PARA EL BIOBANCO

- Sangre
- Orina
- Saliva
- LCR
- L. broncoalveolar
- Biopsias y tejidos sólidos

BIOBANCO. CUESTIONES ÉTICAS

- Altruista
- Niños > 7a deben manifestar además su acuerdo
- Posibilidad de retirar su muestra

- Bioética específica del biobanco
- Investigaciones exclusivamente biomédicas
- Retroinformación escrita al donante/familia
- Confidencialidad

TECNOLOGÍAS REPRODUCTIVAS Y RIESGO DE DEFECTOS CONGÉNITOS

- 2001 Anales de Pediatría (Referee)
- 2012 NEJM Adelaide study congenital defects.

Nacimientos concepción asistida: 6163; Defectos congénitos (5a): 513

Nacimientos sin concep, asistida:302811; Defectos congénitos (5a): 17546

	OR	IC 95%
FIV	1.47	1.33 – 1.62
Esperma intracitoplásmico	1.77	1.47 – 2.12

BIOÉTICA. PROBLEMAS CLÍNICOS PENDIENTES

- Problemas reproductivos en adolescentes con cáncer tratado
- Adolescentes y consumo de alcohol
- (Pseudo) ensayos clínicos
- Soporte vital del pretérmino
- Soporte para el embarazo de adolescentes
- Inmigración

BIOÉTICA PROBLEMAS GENERALES

- **Esquivando el código de la WHO**
- Pseudociencia
- Monetarismo



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Perspective
MARCH 22, 2012

Industry Support of CME — Are We at the Tipping Point?

Michael A. Steinman, M.D., C. Seth Landefeld, M.D., and Robert B. Baron, M.D.

In the 1970s and 1980s, industry-sponsored junkets for physicians, thinly disguised as educational events, were common. Increasing public scrutiny

rounds programs) did not receive any commercial support in 2010.⁵ New rules under “sunshine” provisions of the 2010 health care

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ETICA

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U.S. to Force Drug Firms to Report Money Paid to Doctors

By ROBERT PEAR
Published: January 16, 2012

WASHINGTON — To head off medical conflicts of interest, the Obama administration is poised to require drug companies to disclose the payments they make to doctors for research, consulting, speaking, travel and entertainment.

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EL CONCEPTO DE ÉTICA PUEDE SER CAMBIANTE

XVI Miguel Servet

HOGUERA

XVII Galileo Galilei



(EPPUR SI MUOVE)

Fueron condenados por personas que defendían sus ideas como normales.

MUCHAS GRACIAS