Pesquisa Neonatal para Fibrosis Quística

Situación actual en Argentina y el mundo

7° Congreso Argentino de Neumonología Pediátrica

18-20 Noviembre 2015

Mar del Plata, Buenos Aires

Argentina

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¿Por qué realizar Pesquisa Neonatal en FQ?

- Prevenir y reducir el daño irreversible a nivel pulmonar
- Optimizar el estado nutricional
- Mejorar la calidad de vida

Newborn screening for cystic fibrosis (Review)

2009 The Cochrane Collaboration.

Antecedentes:

- 1979: se describe la elevación y la medición de la TIR en sangre de neonatos con Fibrosis Quística
- La siguiente década la determinación de la TIR se introduce en Australia y algunos países europeos
- 1982: primer programa de pesquisa neonatal para FQ en EEUU .

A Prospective Randomized Trial of Early Diagnosis and Treatment of Cystic Fibrosis: A Unique Ethical Dilemma

NORMAN FOST
PHILIP M. FARRELL

CLIN RES vol 37, no 3, pp 495-500, 1989

Early Diagnosis of Cystic Fibrosis Through Neonatal Screening Prevents Severe Malnutrition and Improves Long-Term Growth

Philip M. Farrell, Michael R. Kosorok, Michael J. Rock, Anita Laxova, Lan Zeng, Hui-Chuan Lai, Gary Hoffman, Ronald H. Laessig, Mark L. Splaingard and the Wisconsin Cystic Fibrosis Neonatal Screening Study Group

Pediatrics 2001;107;1-13

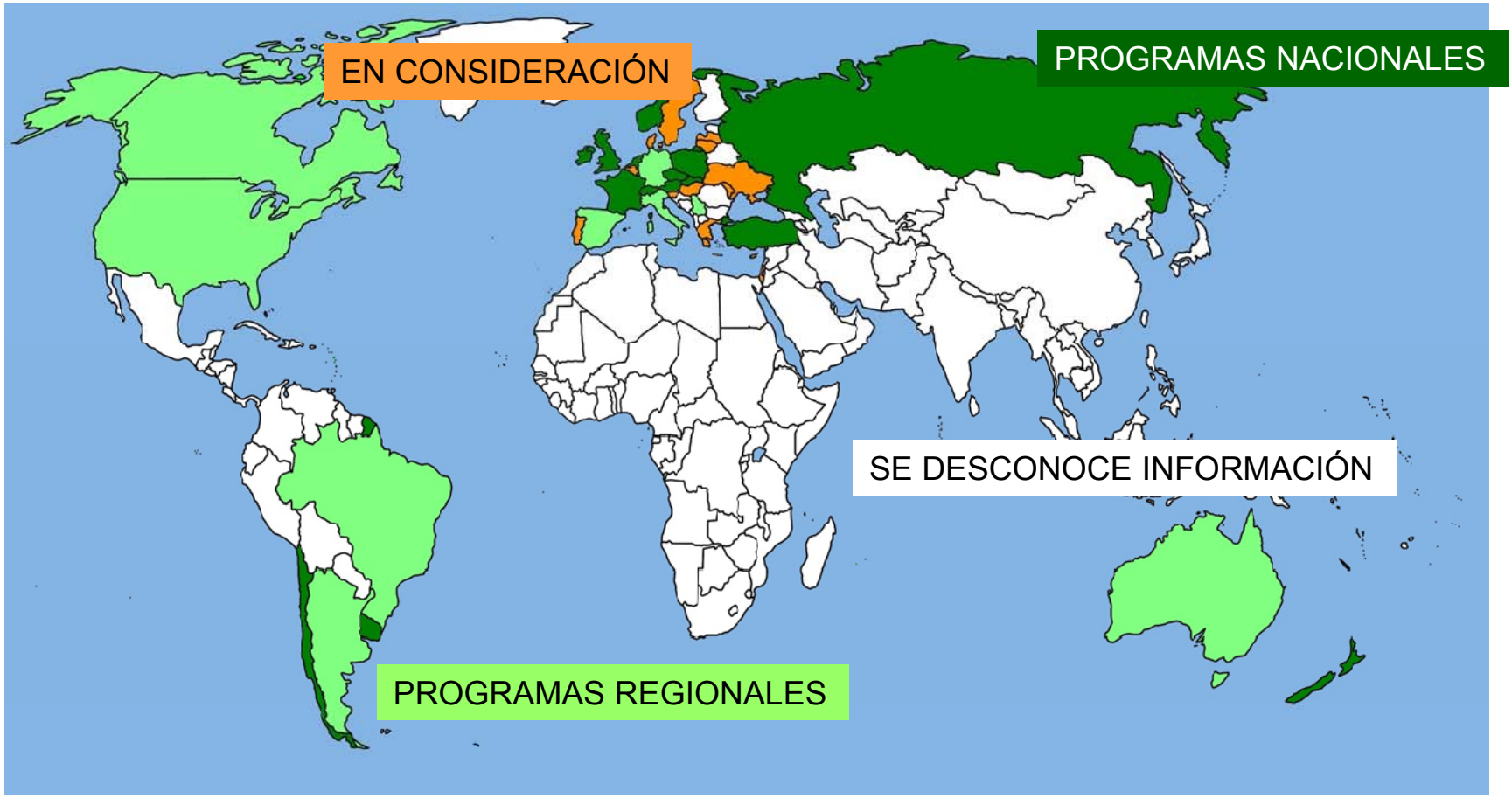
Comparing the Clinical Evolution of Cystic Fibrosis Screened Neonatally to That of Cystic Fibrosis Diagnosed From Clinical Symptoms: A 10-Year Retrospective Study in a French Region (Brittany)

D. Siret, MD,[1]* G. Bretaudeau, MD,[2] B. Branger, MD,[3] A. Dabadie, MD,[2] M. Dagorne, MD,[4] V. David, MD,[1] M. de Braekeleer, MD,[5] V. Moisan-Petit, MD,[6] G. Picherot, MD,[7] G. Rault, MD,[8] V. Storni, MD,[8] and M. Roussey, MD[2]

Pediatric Pulmonology 35:342–349 (2003)

CF NBS 2015

ECFS CF Neonatal Screening Working Group



A survey of newborn screening for cystic fibrosis in Europe

Kevin W. Southern [a], Anne Munck [b], Rodney Pollitt [c], Georges Travert [d], Luisa Zanolla [e], Jeannette Dankert-Roelse [f], Carlo Castellani [g,*]
on behalf of the ECFS CF Neonatal Screening Working Group

Description of NBS programmes included in survey

	Area	2nd tier	3rd tier	4th tier	Details
Two-tier protocols					
I	Liguria	ST	–	–	
Three-tier protocols					
CZ	Western Czech republic	MUT	ST	–	PS
UK	Wales	MUT	ST	–	
UK	Northern Ireland	IRT-2	ST	–	
UK	Leeds Halifax Jersey	MUT IRT-2	ST	–	
I	Emilia Romagna	IRT-2	ST	–	
I	Calabria	IRT-2	ST MUT	–	PS
I	Sardinia	ST	MUT	–	PS
I	Lombardy	MUT IRT-2	ST	–	IR
I	Marche	MUT IRT-2	ST	–	PS
I	Tuscany	MP IRT-2	ST	–	
I	Piedmont	MUT IRT-2	ST	–	PS
I	Lazio 1	IRT-2	ST MUT	–	
I	Lazio 2, Umbria	MUT IRT-2	ST	–	
I	Western Sicily	MUT IRT-2	ST	–	
A	Austria	IRT-2	ST	–	
SP	Catalunya	IRT-2	MUT ST	–	PS
SP	Castilla-Leon	IRT-2 MUT	ST		
SP	Galice	IRT-2	MUT ST	–	PS
Four-tier protocols					
F	France	MUT	IRT-2	ST	IRT-2 if MUT-tive
PL	Poland	MUT	IRT-2	ST	PS 1999–2003; IRT-2 if MUT-tive
UK	South Yorkshire East Midlands	MUT	IRT-2	ST	IRT-2 if 1 mutation or IRT-1 >99.9th centile
UK	Scotland	MUT	IRT-2	ST	IRT-2 if 1 mutation or no mutations and non-Caucasian
UK	Northamptonshire	MUT	IRT-2	ST	IRT-2 if 1 or no mutations
UK	East Anglia	MUT	IRT-2	ST	IRT-2 if 1 mutation or IRT-1 >99.9th centile
I	Veneto Trentino Alto-Adige	MUT MP	IRT-2	ST	IRT-2 if MUT and MP-tive and IRT-1 twice the cutoff

- 26 Centros (UK, Francia, Italia, España, Austria, Polonia, y Republica Checa),entre 2004 – 2005.
- 100 % 1º TIR
- 2 Centros: TIR/TS
- 7 Centros : TIR/TIR
- 19 TIR /ADN (73%). (31 mutaciones: cobertura ≥ 83%)

Journal of Cystic Fibrosis 6 (2007) 57 – 65

A survey of newborn screening for cystic fibrosis in Europe

Kevin W. Southern [a], Anne Munck [b], Rodney Pollitt [c], Georges Travert [d], Luisa Zanolla [e], Jeannette Dankert-Roelse [f], Carlo Castellani [g,*]
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Number of newborns screened and case recognition

Areas		Newborns screened per year	CF incidence (calculated)	Age at diagnosis (weeks)
Two-tier protocols				
I	Liguria	11,000	1/4400	8–9
Three-tier protocols				
CZ	Western Czech republic *	45,500	1/9100	4–6
UK	Wales	32,500	1/2700	<4
UK	Northern Ireland	23,000	1/2850	4–6
UK	Leeds Halifax Jersey	11,000	1/2750	3–6
I	Emilia Romagna	33,000	1/4700	8–9
I	Calabria	16,000	–	6–9
I	Sardinia	14,000	–	17
I	Lombardy	92,000	1/4600	3–5
I	Marche	13,000	1/5200	8–9
I	Tuscany	30,000	1/3500	6
I	Piedmont	37,000	1/2650	6
I	Lazio 1	28,500	1/3150	NA
I	Lazio 2, Umbria	33,000	NA	NA
I	Western Sicily	20,000	1/2500	6
A	Austria	80,000	1/3500	5–6
SP	Catalunya	62,500	1/5700	7–10
SP	Castilla-Leon	18,000	1/4000	3–12
SP	Galice	21,000	1/10,500	4–5
Four-tier protocols				
F	France	800,000	1/4700	5
PL	Poland	90,000	1/5000	4–6
UK	South Yorkshire East Midlands	55,000	1/2450	NA
UK	Scotland	54,000	1/2700	From 3 upwards
UK	Northamptonshire	8000	1/2250	3–8
UK	East Anglia	25,000	1/2800	3–6
I	Veneto Trentino Alto-Adige	52,000	1/4150	3–6

Combined results (presented as median and interquartile ranges)	
Years of screening	7 (3.6–18.8)
Screened per year	30,000 (18,000–54,000)
IRT +ive per year	295 (148–825)
Sweat tests per year (as part of programme)	70 (20–129)
Number of CF cases per year	9 (5–14)
Carriers per year	17 (13–25)
False negatives since start of programme	2 (1–5)
False negatives per year	0.3
False negatives reported?	Yes: 15 (57.7%) No: 5 (19.2%) Not always: 2 (7.7%)
Causes of false negatives	First IRT low: 17 (65.4%) No mutations recognised: 1 (3.9%) Second IRT low: 4 (15.4%) Negative sweat test: 1 (3.9%)

- Pesquisados : 18000 – 54000 niños/año.
- La incidencia varia desde 1. 2250 a 10. 500 (1: 3500).
- Media de 9 pacientes / año.

Journal of Cystic Fibrosis 6 (2007) 57–65

The prevalence of cystic fibrosis in the European Union

Philip M. Farrell

Population and prevalence of patients with CF in E.U. countries

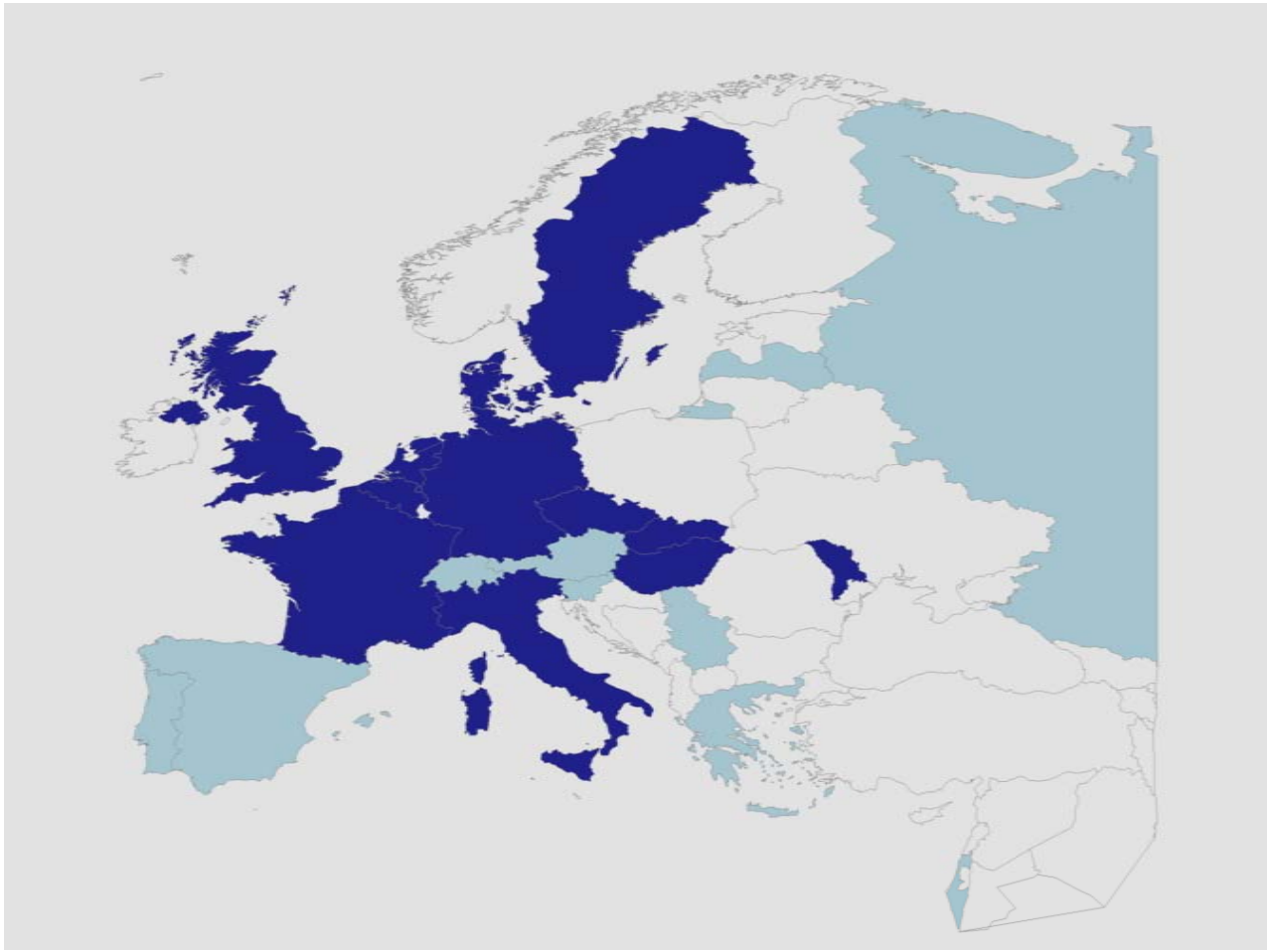
	Population in 2004 (thousands)	# CF patients	CF prevalence (per 10,000)	Estimated CF incidence	Source(s)
Austria	8,175	686	0.839	1:3500	Ia, [1]
Belgium	10,348	1065	1.03	1:2850	Ib, IIa, [13,14]
Bulgaria	7,518	170	0.226	1:2500	[13]
Cyprus	776	26	0.335	1:7914	[15]
Czech Republic	10,246	570	0.556	1:2833	Ic, IIa, [14,16]
Denmark	5,413	412	0.761	1:4700	IIa, [14,17,18]
Estonia	1,342	83	0.618	1:4500	[17]
Finland	5,215	64	0.123	1:25000	Id, [17,19]
France	60,424	4533	0.750	1:4700	Ie, IIa, IIe, [1]
Germany	82,425	6835 [a]	0.829 [a]	1:3300	If, IIa, [14,16,20]
Greece	10,648	555	0.521	1:3500	Ig, [14]
Hungary	10,032	410	0.409		Ih
Ireland	3,970	1182	2.98	1:1353	Ii, IIb, [8]
Italy	58,057	5064	0.872	1:4238	IIc, [21]
Latvia	2,306	24	0.104		[7]
Lithuania	3,608	47	0.130		[7]
Luxembourg	463	20	0.431		[7]
Malta	397	23	0.579		IV
Netherlands	16,318	1275	0.781	1:4750	IIa, [22]
Poland	38,580	987	0.256	1:5000	Ij, [1]
Portugal	10,524	285 [a]	0.271 [a]	1:6000	Ik,[7]
Romania	22,356	238	0.106	1:2056	[23]
Slovakia	5,424	340	0.627	1:1800	IIa, [24]
Slovenia	2,011	66	0.328	1:3000	[7,25]
Spain	40,281	2200 [a]	0.546 [a]	1:3750	Il, [13,14]
Sweden	8,986	362	0.403	1:5600	IIa, [26]
United Kingdom	60,271	8284	1.37	1:2381	Im, IIa, IId, [9]

Journal of Cystic Fibrosis 7 (2008) 450–453

ECFSPR

European Cystic Fibrosis Society Patient Registry

Annual data report (year 2010)
Version 01.2014



Países que mandan datos como Registro Nacional

Países con registros propios que envían datos

ECFS Patient Registry

Proportion of patients who underwent neonatal screening, by country and overall. Patients 5 years old or younger seen in 2010.

2010 data

55% de los niños de 5 años o menores fueron diagnosticados por Pesquisa neonatal

Initial evaluation of a biochemical cystic fibrosis newborn screening by sequential analysis of immunoreactive trypsinogen and pancreatitis-associated protein (IRT/PAP) as a strategy that does not involve DNA testing in a Northern European population

Olaf Sommerburg · Martin Lindner · Martina Muckenthaler · Dirk Kohlmueller
Svenja Leible · Reinhard Feneberg · Andreas E. Kulozik · Marcus A. Mall ·
Georg F. Hoffmann

J Inherit Metab Dis 2010; 33:S263

Moving towards a national newborn screening programme for CF in Germany Olaf Sommerburg, University Children's Hospital III, Heidelberg, Germany

The Neonatal Screening Working Group Annual Meeting Brussels, Belgium 10th June 2015

Desde el 2008 dos centros regionales de Alemania implementaron el algoritmo **TIR/PAP**, como protocolo de pesquisa para FQ

2013 CF Fundation Patient Registry Annual Data Report

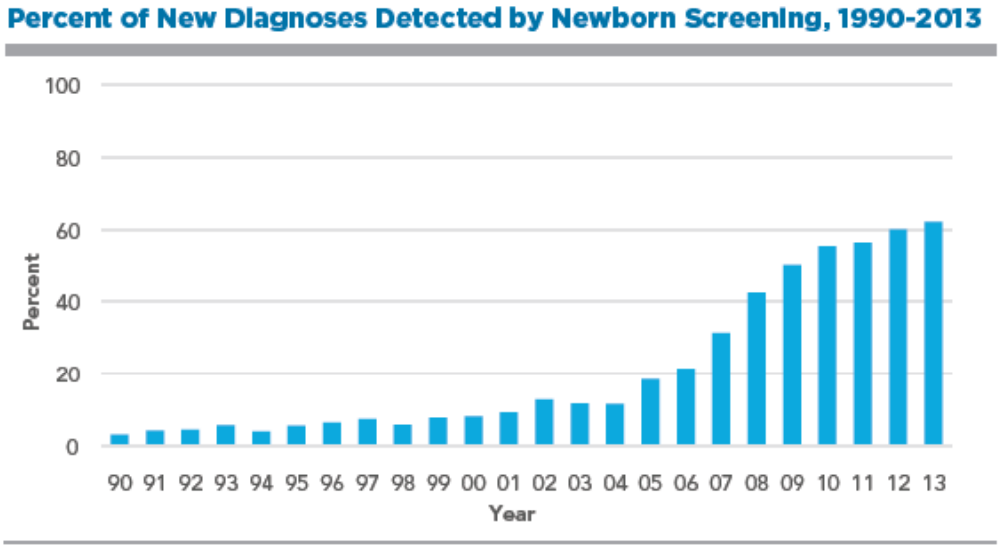
Philip Farrell, University of Wisconsin-Madison, described the last ten years of NBS for CF in the US, moving from four States offering screening in 2004 to total coverage by 2009. Each State organizes their own programme, leading to some variability across the country, however there are essentially three types of protocol. The majority of States use an IRT-DNA protocol. In six States a second dried blood sample is routinely collected at 2-3 weeks and DNA analysis only undertaken with persistent raised IRT. Finally eleven States (including Alaska and Hawaii) use an IRT-IRT protocol with no DNA analysis. One State (California) employs extended DNA analysis if one mutation is identified on the initial limited panel. With successful implementation of NBS across the US, the emphasis has now turned to quality improvement and Professor

The Neonatal Screening Working Group Annual Meeting , October 2013

IRT/ADN (mayoría de los estados)

IRT/ADN EXTENDIDO (California)

IRT/IRT (11 estados incluido Alaska y Hawaii)



Pesquisa Neonatal en América Latina: Historia y Realidad

- 1973 (México). Antonio Velásquez inicia protocolo para fenilcetonuria
- 1976 (Brasil). Benjamín Schmidt funda el Laboratorio para detectar “Errores Congénitos del Metabolismo”
- 1994 (México). Primer programa estructurado con la Salud Pública
- 1986 – 1995: Cuba, Argentina, Brasil, Chile, Colombia, Costa Rica, México, Uruguay y Venezuela comienzan con Programas de Pesquisa

América Latina: Región de Enormes Diferencias

Comparación entre los Programas de Pesquisa Latinoamericanos

LATINOAMERICA	% COBERTURA	QUE PESQUISAN
CHILE	98	HC, PKU, MASA (selectivo)
COSTA RICA	99.3	HSC, HC, MASA (24 enf.), FQ (ad)
CUBA	99.5	HSC, HC, PKU, GAL
URUGUAY	99.5	HC, PKU, HSC, FQ, MASA (PP)
BRASIL	80.2	HC, PKU, Hb, HSC(ad), GAL(ad) MSUD(ad)
MEXICO	70	HC, PKU(ad), HSC(ad), MSUD(ad)
ARGENTINA	85	HC, PKU, HSC, GAL, MSUD, FQ, BIOTINIDASA
COLOMBIA	80	HC, PKU(ad), HSC(ad), GAL(ad)
PANAMÁ	48	HC, G6PDH, HSC, GAL, PKU, Hb
PARAGUAY	30	HC, PKU, FQ, HSC(ad), FQ(ad)
VENEZUELA	25-30	HC, PKU, HSC(ad), GAL(ad)
NICARAGUA	6	HC
PERÚ	10	HC, HSC
BOLIVIA	A demanda	
GUATEMALA	A demanda	
ECUADOR	A demanda	
REPUBLICA DOMINICANA	A demanda	
EL SALVADOR	0	
HONDURAS	0	
HAITÍ	0	

Grupo I: Cuba, Costa Rica, Chile y Uruguay (100% de cobertura)

Grupo II: Brasil, México y Argentina (60 a 80% de cobertura)

Grupo III: Colombia, Paraguay y Venezuela (< 30% de cobertura)

Grupo IV: Nicaragua y Perú (4 a 6% de cobertura)

Grupo V: Guatemala, Rep. Dominicana, Bolivia, Panamá y Ecuador (1% de cobertura)

Grupo VI: El Salvador, Honduras y Haití (sin cobertura)

Pesquisa Neonatal en América Latina: Historia y Realidad

- En la actualidad existen países que carecen de Programas de Pesquisa Neonatal
  - Falta de recursos económicos
  - Presencia de otras prioridades sanitarias
  - Falta de apoyo de las autoridades sanitarias
  - Retraso en la implementación de los programas
  - Dificultades en la integración de los programas con las políticas de salud

América Latina: Región de Enormes Diferencias

PAIS	ALGORITMO	INCIDENCIA
BRASIL	IRT/IRT	1:6.551 in Santa Catarina 1:9.089 in Paraná 1:10.840 in Minas Gerais
COLOMBIA	IRT/DNA	2011: 1:8297
URUGUAY	IRT/PAP	1:2500- 3000
CHILE	IRT/IRT IRT/PAP	1:8000
MEXICO	IRT/IRT	2002: 1: 8500
COSTA RICA	IRT/IRT	1:5000
ARGENTINA	IRT/IRT	1: 6100 a 1: 7500

Fernando Abreu da Silva, Francisco Caldeira Reis, Luiz Vicente Ribeiro Ferreira da Silva Filho

José Luis Lezana Fernandez,

Graciela Queiruga. Mem. Inst. Investig. Cienc. Salud.2011;9:72-77, Maria Catalina Pinchak Rosales,

Catalina Vasquez Sagra,

Jose Pablo Gutierrez Schwanhauser,

Estrategias de Pesquisa Neonatal para FQ: Ventajas y Desventajas

	IRT/IRT	IRT/DNA	IRT/DNA/IRT	IRT/IRT/DNA	IRT/PAP
Especificidad	<	>>	>>	>	>
Sensibilidad	<	>	>	<	>
VPP	<	>>	>>	>	>
Aplicabilidad	General	Relativa	Relativa	Relativa	General
Necesidad de 2° muestra	Si	No	Si, < proporc.	Si	No
Detección portadores	No	Si	Si	Si	No
Detección formas leves	No	Si	Si	Si	No
Asesoramiento genético	Si	Amplio	Amplio	Si	Si
Tiempo para diagnóstico	>	<<	<	>	<<
Pedidos Test del Sudor	+++	++	+	++	+
Costos	+	+/+++	++/+++	++	+

G Borrajo. 2006

CFTR gene analysis in Latin American CF patients: Heterogeneous origin and distribution of mutations across the continent

A total of 4354 unrelated CF chromosomes were studied from 10 countries. The results show a wide distribution of 89 different mutations, with a maximum coverage of 62.8% of CF chromosomes/alleles in the patient's sample. Most of these mutations are frequent in Spain, Italy, and Portugal, consistent with the origin of the European settlers. A few African mutations are also present in those countries which were part of the slave trade.

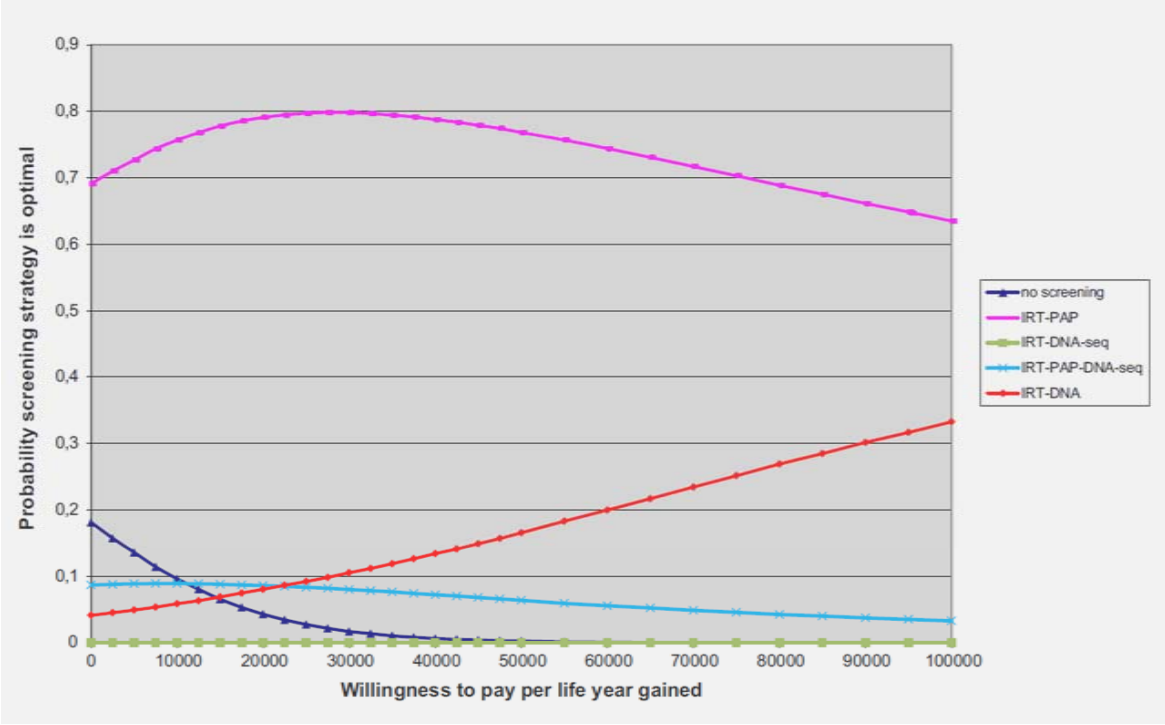
Country	Chromosomes analysed	p.F508del		p.G542X		p.N1303K		p.W1282X		p.R1162X		Unknown	
		n	%	*n*	%	*n*	%	*n*	%	*n*	%	*n*	%
Argentina	1246	737	59.15	61	4.90	28	2.25	27	2.17	9	0.72	271	21.75
Brazil	1858	800	43.06	99	5.33	34	1.83	11	0.59	25	1.35	789	42.46
Chile	252	99	39.28	14	5.55	0	0.00	8	3.17	3	1.19	115	45.63
Colombia	218	90	41.28	9	4.13	2	0.92	3	1.38	2	0.92	84	38.53
Costa Rica	48	11	22.92	12	25.00	–	–	–	–	–	–	25	52.08
Cuba	144	49	34.03	–	–	–	–	–	–	–	–	95	65.97
Ecuador	102	32	31.37	2	1.96	1	0.98	–	–	–	–	65	63.72
Mexico	274	115	41.97	16	5.84	5	1.82	–	–	–	–	88	32.11
Uruguay	76	43	56.58	6	7.89	2	2.63	–	–	3	3.95	11	14.47
Venezuela	136	57	41.91	2	1.47	–	–	–	–	–	–	77	56.62
Total	4354	2033	46.69	221	5.08	72	1.65	49	1.13	42	0.96	1620	37.21

Conclusion: The profile of mutations in the CFTR gene, which reflects the heterogeneity of its inhabitants, shows the complexity of the molecular diagnosis of CF mutations in most of the Latin American countries.

MM Pérez et al. Journal of Cystic Fibrosis.2007;6:194–208

Cost-effectiveness of newborn screening for cystic fibrosis determined with real-life data

Was used primary data to compare cost-effectiveness of four screening strategies for NBSCF: IRT–PAP, IRT–DNA, IRT–DNA–sequencing, and IRT–PAP–DNA–sequencing, each compared to no-screening.



Conclusion: NBSCF is as an economically justifiable public health initiative. Of the four strategies tested IRT–PAP is the most economic and this finding should be included in any decision making model, when considering implementation of newborn screening for CF.

CPB van der Ploeg, et al. J Cyst Fibros (2014)

Fortalecimiento de la Detección Precoz de Enfermedades Congénitas

Objetivos

- Fortalecer los programas provinciales de pesquisa neonatal.
- Promover que la pesquisa alcance la cobertura del 100% de los recién nacidos vivos del sector público.
- Contribuir al seguimiento clínico de todos aquellos niños detectados como positivos confirmando su diagnostico.
- Colaborar en el tratamiento de los casos detectados como positivos en forma precoz.

Programa de Pesquisa Neonatal en FQ Epidemiología en Argentina

- Un análisis realizado por el Programa Materno Infantil de la Nación sobre Pesquisa Neonatal reveló que el 74% de las provincias realizan algún tipo de pesquisa y el 34% de las provincias que efectúan pesquisa neonatal la realizan sólo en un 50 % de la población objetivo.

- En el segundo semestre de 2012 se firmó un convenio entre la Sociedad Argentina de Pediatría (Comité Nacional de Neumonología) y el Instituto Nacional de Enfermedades Respiratorias (INER/ANLIS) para la implementación de un Registro Nacional de pacientes y se comenzó la carga de datos, de carácter voluntario

- Pesquisa Neonatal: 28.8 %,

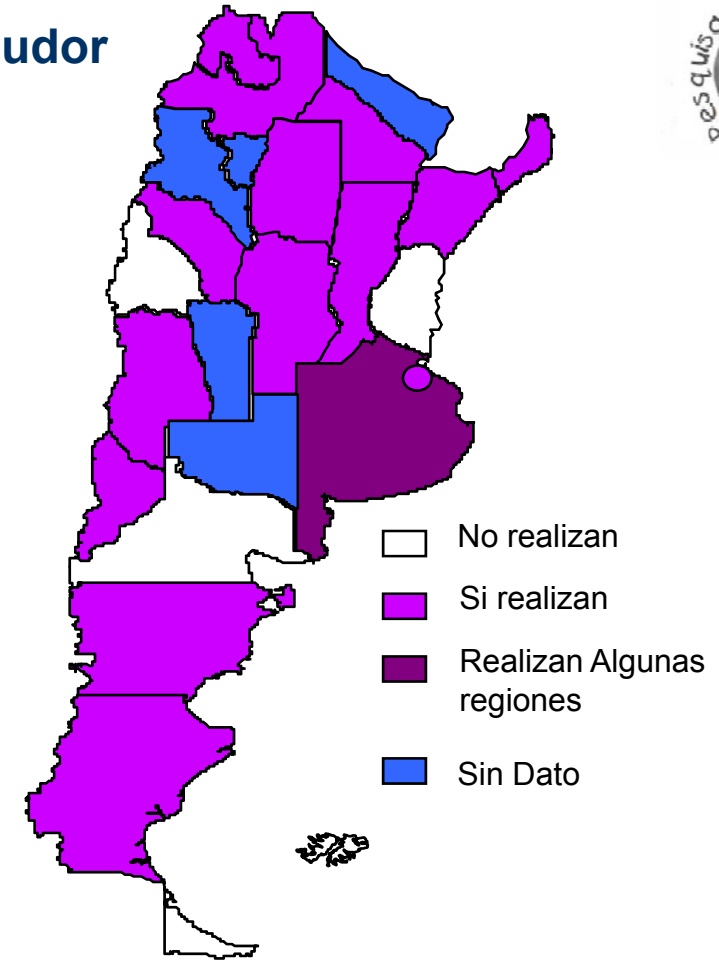
Grupo Registro Nacional de Fibrosis Quística – ARGENTINA

2014

Programa Nacional de Pesquisa Neonatal en FQ

TIR

Test Sudor



Pesquisa Neonatal

Dirección Nacional de Maternidad e Infancia

Ministerio de Salud
Presidencia de la Nación

Antecedentes:

- *Fenilcetonuria*

  Ley 23413 sancionada el 10 de octubre de 1986

- *Hipotiroidismo congénito*

  Ley 23874 sancionada el 28 de septiembre de 1990

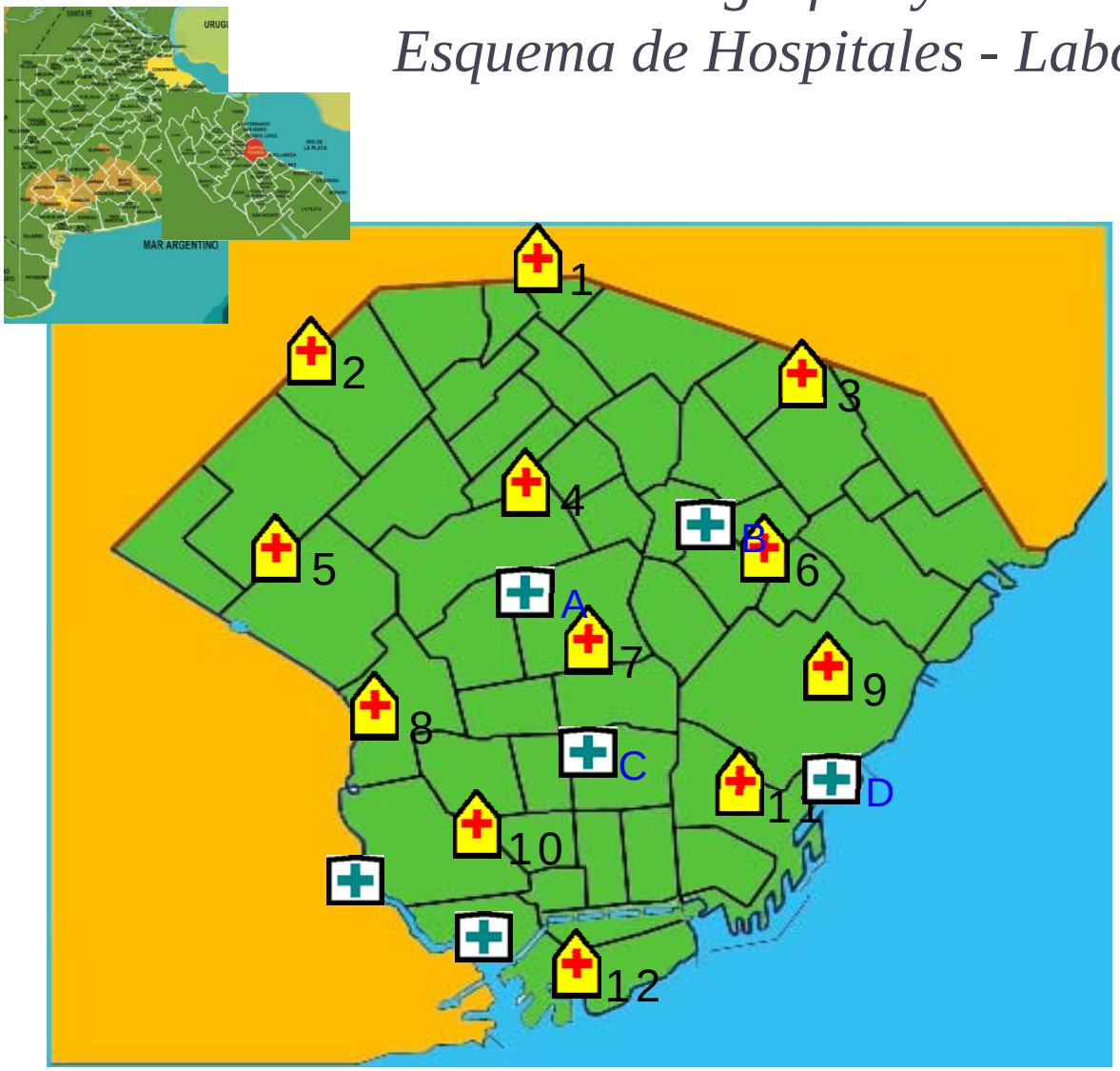
- ***Fibrosis Quística***

  Ley 24438 sancionada el 21 de diciembre de 1994

Programa de Pesquisa Neonatal del Gobierno de la Ciudad de Buenos Aires

- 2002– Pesquisa Fibrosis Quística

Distribución Geográfica y Circuito Analítico: Esquema de Hospitales - Laboratorios



MATERNIDADES

1. Vélez Sarfield
2. Santojanni
3. Pirovano
4. Álvarez
5. Piñero
6. Sardá
7. Durand
8. Penna
9. Rivadavia
10. Ramos Mejía
11. Fernández
12. Argerich

LABORATORIOS

A. Garrahan
B. Durand
C. Gutiérrez
D. Ramos Mejía

CENTRO DE CONFIRMACION DIAGNOSTICA

Centro Respiratorio Hospital de Niños “R Gutiérrez”

Programa de Pesquisa Neonatal para FQ del Gobierno de la Ciudad de Buenos Aires

PESQUISA

TIR ≥ 60 ng/ml

↓

Se repite 2°TIR antes de los 25 días de vida

↓

2° TIR ≥ 60 ng/ml

DIAGNÓSTICO

TEST del SUDOR → POSITIVO o DUDOSO →

- Htal Ricardo Gutiérrez
- Htal Pedro Elizalde

↓

ESTUDIO GENETICO
32 mutaciones

TEST del SUDOR

↓

NORMAL

↓

ALTA

- Período evaluado : 145 meses (Dic 02- Dic 14)
- Nº de Nacimientos : 364782

TIR elevada
1462 (0.4 %)

Test del Sudor

FQ: 49

Incidencia 1: 7444

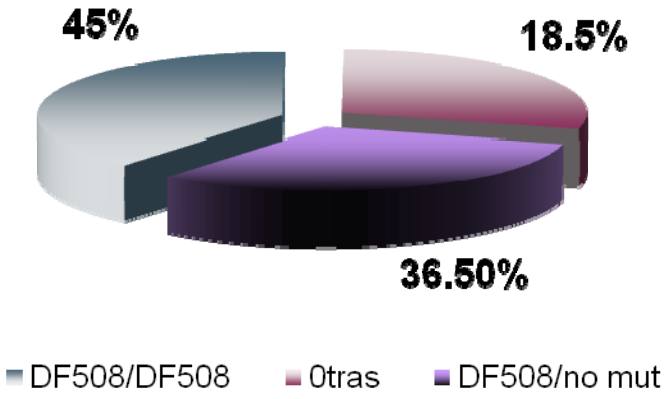
Estudio Genético

P 508 del homocigota: 20

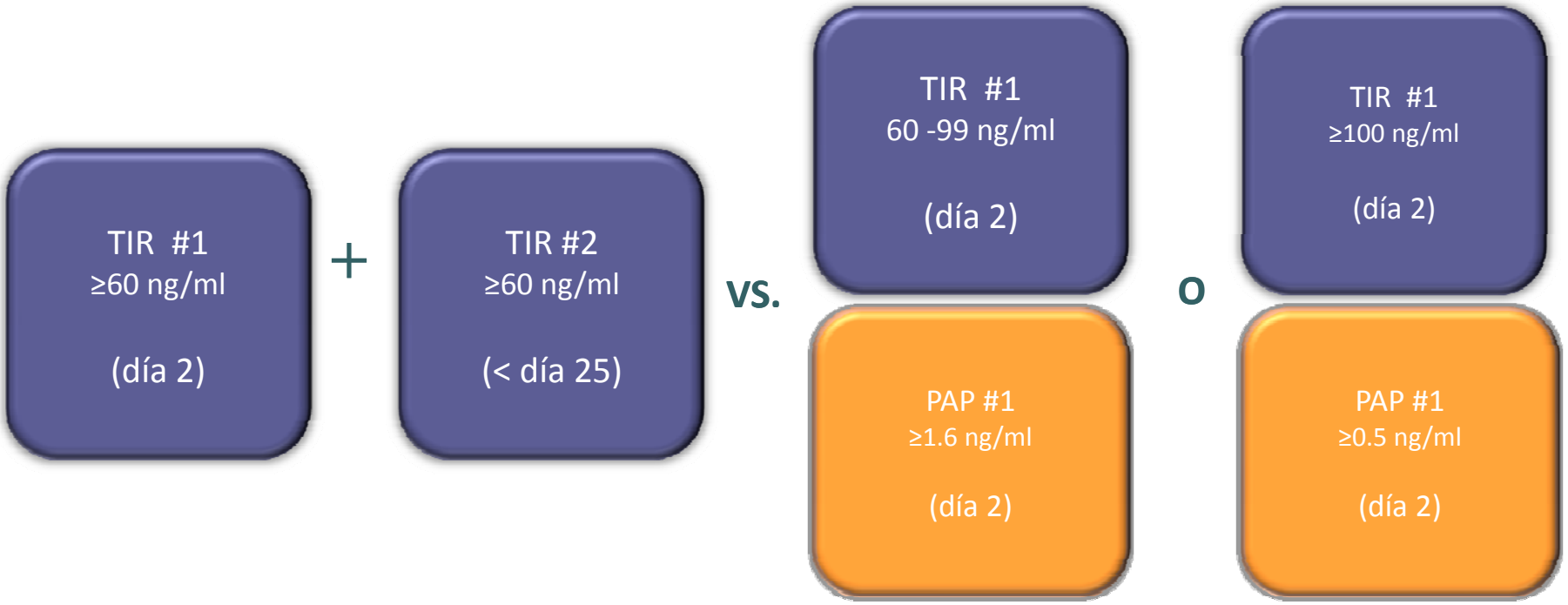
P 508 del heterocigota: 16

Otras: 13

Estudio genético



Comparación de dos Estrategias de Pesquisa Neonatal para Fibrosis Quística: Estudio Piloto de la Ciudad Autónoma de Buenos Aires



La confirmación diagnóstica se efectuó por Test del Sudor

N : 15000

1º TIR anormal: 105 (0.7)

2º TIR anormal : 83

A Teper et al. ECFS, Gothenburg 2014

Comparación de dos Estrategias de Pesquisa Neonatal para Fibrosis Quística: Estudio Piloto de la Ciudad Autónoma de Buenos Aires

TIR 2 Anormal	**20 (24 %)** **Sensibilidad:** 100% (95% CI: [2.5% - 100%]) **Especificidad:** 76.8% (95% CI: [66.2% - 85.4%]) **VPP:** 5% (95% CI: [0.1% - 24.9%]) **VPN:** 100% (95% CI: [94.3% - 100%])
PAP Anormal (1^{era} muestra)	**6 (7.2%)** **Sensibilidad:** 100% (95% CI: [2.5% - 100%]) **Especificidad:** 93.9% (95% CI: [86.3% - 98%]) **VPP:** 16.7% (95% CI: [0.4% - 64.1%]) **VPN:** 100% (95% CI: [95.3% - 100%])
Test del Sudor Anormal	1 (FQ) TIR/TIR: 190/147 ng/ml PAP: 1.95 ng/ml
Estudio Genético	1 ΔF508 homocigota (FQ)
	1 G542X heterocigota (portador) TIR/TIR: anormal PAP: normal TS: normal

CONCLUSIÓN: *la estrategia TIR/PAP permite disminuir la recitación de pacientes para segunda muestra de TIR, sin modificar la sensibilidad y especificidad de la pesquisa neonatal.*

A Teper et al. ECFS, Gothenburg 2014

Programa de Pesquisa Neonatal para FQ del Gobierno de la Ciudad de Buenos Aires, (2015)

PESQUISA

TIR ≥ 50 ng/ml + **PAP (TIR ≥ 100=0.5ng/ml -TIR ≥50 =1.6 ng/ml)**

↓

Se repite 2°TIR antes de los 25 días de vida

↓

2° TIR ≥ 50 ng/ml

PESQUISA

DIAGNÓSTICO

TEST del SUDOR → POSITIVO o DUDOSO →

- Htal Ricardo Gutiérrez
- Htal Pedro Elizalde

↓

ESTUDIO GENETICO
32 mutaciones

TEST del SUDOR

↓

NORMAL

↓

ALTA

New Challenges in the Diagnosis and Management of Cystic Fibrosis

Hara Levy, MD
Philip M. Farrell, MD, PhD

CFTR-RD

- CBAVD
- Pancreatitis aguda recurrente o crónica
- Bronquiectasias diseminadas

Journal of Cystic Fibrosis Volume 10 Suppl 2 (2011) S86–S102

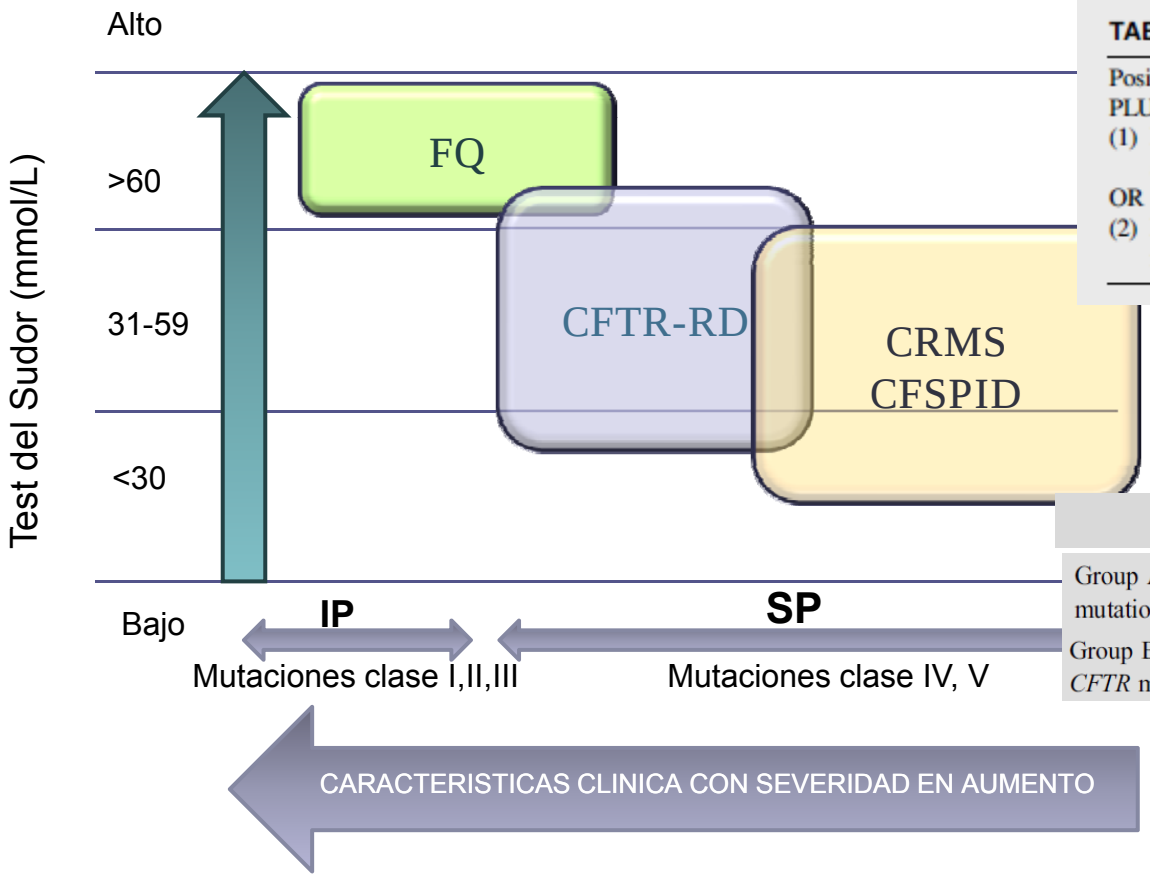


TABLE 2—Criteria for Diagnosis of CRMS[5]

Positive CF NBS test
PLUS
(1) Intermediate sweat chloride concentrations on at least 2 occaisions and fewer than 2 CF-causing CFTR gene mutations
OR
(2) Normal sweat chloride and 2 CFTR gene mutations, of which no more than 1 is known to be CF-causing.

Pediatric Pulmonology 46:1079–1084 (2011)

CFSPID

Group A, normal sweat chloride value (<30 mmol L^{-1}) and two *CFTR* mutations, at least one of which has unclear phenotypic consequences

Group B, intermediate sweat chloride value (30–59 mmol L^{-1}) and one or no *CFTR* mutations

Journal of Cystic Fibrosis xx (2015) xxx – xxx

J Pediatr. 2015 June ; 166(6): 1337–1341

Resumen

Objetivos del programa de pesquisa neonatal

- Oportunidad temprana
- Tratamiento y cuidado especial
- Mantener normal crecimiento y desarrollo
- Retrasar complicaciones pulmonares y deterioro funcional

Resumen

- Brasil, Argentina , Costa Rica y Uruguay poseen programas nacionales de Pesquisa Neonatal para Fibrosis Quística
- La incidencia en la región varía desde 1: 7000 a 1: 10000
- Incidencia de Fibrosis Quística en CABA : 1: 7444
- La utilización del protocolo TIR/PAP optimizaría la estrategia de pesquisa
- Desafío: método “óptimo” para cada población