Autismo Bizkaia
APNABI

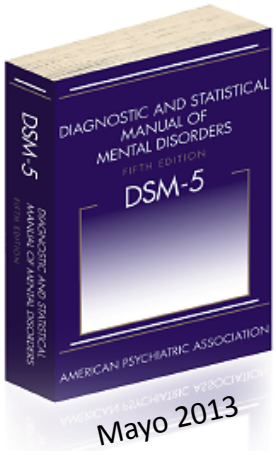
LA INVESTIGACIÓN EN EL AUTISMO

Joaquín Fuentes Biggi
Donostia / San Sebastián

POLICLINICA GIPUZKOA

GAUTENA

DSM-5: Trastorno del Espectro del Autismo



Inicio Temprano
Deterioro clínico significativo

Edad
Nivel de Inteligencia
Dominio del Lenguaje
Asociado a otros trastornos

DSM5: Trastorno del Espectro del Autismo

A. Deficiencias en la comunicación social y en la interacción social en entornos diversos

Deficiencias en todas las 3 áreas:

1. Reciprocidad socio-emocional
2. Comportamientos comunicativos no-verbales utilizados para la interacción social
3. Desarrollar y mantener relaciones adecuadas al nivel de desarrollo

DSM5: Trastorno del Espectro del Autismo

B. Patrones restrictivos, repetitivos, de conductas, intereses o actividades

Deficiencias en al menos 2 áreas:

1. Uso estereotipado o repetitivo del habla, los movimientos motrices o los objetos
2. Adhesión excesiva a las rutinas, patrones ritualizados o excesiva resistencia al cambio
3. Intereses fijos,marcadamente restringidos
4. Hiper- o hipo-reactividad a los estímulos sensoriales

Las clasificaciones no clasifican a las personas

La intervención no viene determinada por las características *grupales* de esta población, sino por las características *específicas* de cada individuo

PERSONALIZA y CONTEXTUALIZA!!

Identified Prevalence of Autism Spectrum Disorder

ADDM Network 2000-2010
Combining Data from All Sites

Surveillance Year	Birth Year	Number of ADDM Sites Reporting	Prevalence per 1,000 Children (Range)	This is about 1 in X children...
2000	1992	6	6.7 (4.5 – 9.9)	1 in 150
2002	1994	14	6.6 (3.3 – 10.6)	1 in 150
2004	1996	8	8.0 (4.6 – 9.8)	1 in 125
2006	1998	11	9.0 (4.2 – 12.1)	1 in 110
2008	2000	14	11.3 (4.8 – 21.2)	1 in 88
2010	2002	11	14.7 (14.3 – 15.1)	1 in 68

1 / 42 ♂

1 / 189 ♀

IQ <70 [**31%**]
IQ 71-84 [**22%**]
IQ >85 [**46%**]

Fuentes J et al. *Autism spectrum disorder*. In Rey JM (ed), *IACAPAP e-Textbook of Child and Adolescent Mental Health*. Geneva: International Association for Child and Adolescent Psychiatry and Allied Professions 2014.

Prevalencia de Autismo en 55.000 escolares de 7 a 12 años en Corea del Sur: 2,64% (1 de cada 38)

Nueva distribución por sexo en esta muestra: 2.5 – 5 / 1

Kim YS et al. (2011) J Am Acad Child Adolesc Psychiatry, 168:904-12
Kim YS et al. (2014) J Am Acad Child Adolesc Psychiatry. 53:500-8

1

EL AUTISMO EMPIEZA ANTES DE NACER (“Pedazos”)

11 TEA: 10 tenían “pedazos” de desarrollo anormal (2º trimestre)
11 NO-TEA: 1 tenía “pedazos” de desarrollo anormal

+ Frontal (comunicación y social), + Temporal (lenguaje), - Visual

R. Stoner. *NJM*, March 2014

2

TEA: Dos veces más riesgo en reproducción asistida

Pero las diferencias disminuyeron enormemente si la madre era menor de 34 años y **NO HABÍA COMPLICACIONES O EMBARAZOS MÚLTIPLES**

C. Fountain. *Public Health*, March 2015

3

Potenciales marcadores para el diagnóstico y tratamiento

Expresión de RNA en niños jóvenes y su efecto en proteínas relacionadas con la inflamación y la inmunidad ("Firma")

ACIERTO EN EL 82% DE LOS CASOS DE TEA

vs.

Controles, Retrasos Globales o Problemas de Lenguaje

87 con TEA vs. 55 sin TEA (1 – 4 años)

T. Pramparo. *JAMA*, March 2015

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MSSNG (Google + Autism Speaks)

SECUENCIACIÓN DEL GENOMA COMPLETO DE 10.000 FAMILIAS CON AUTISMO Y UBICACIÓN EN LA "NUBE" PARA LA INVESTIGACIÓN MUNDIAL

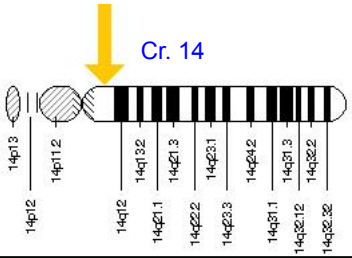
Hopital for Sick Children, Toronto

5

MUTACIÓN GENÉTICA PARA UN SUBTIPO ESPECÍFICO

Frente amplia, ojos separados, estreñimiento, problemas del sueño y TEA

Mutación del Gen CHD8



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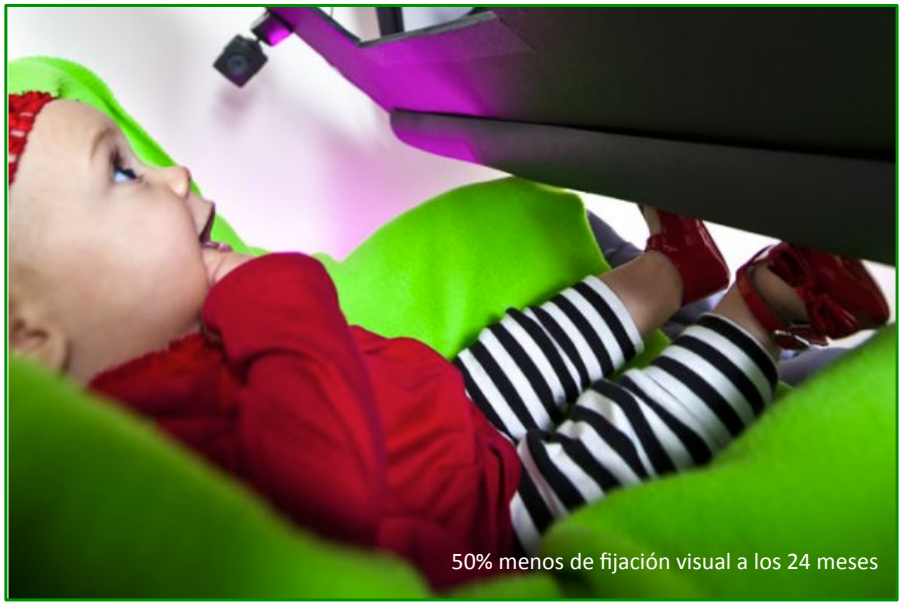
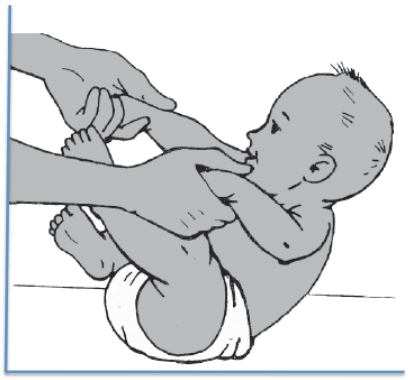


Jones W & Klin A. Attention to eyes is present but in decline in 2-6-month-old infants later diagnosed with autism. 2013 *Nature*, 504: 427-31

7



El retraso en el sostén cefálico precede en muchos casos a la manifestación de síntomas de autismo a los 6 meses de edad

Fuentes J et al. *Autism spectrum disorder*. In Rey JM (ed), *IACAPAP e-Textbook of Child and Adolescent Mental Health*. Geneva: International Association for Child and Adolescent Psychiatry and Allied Professions 2014.

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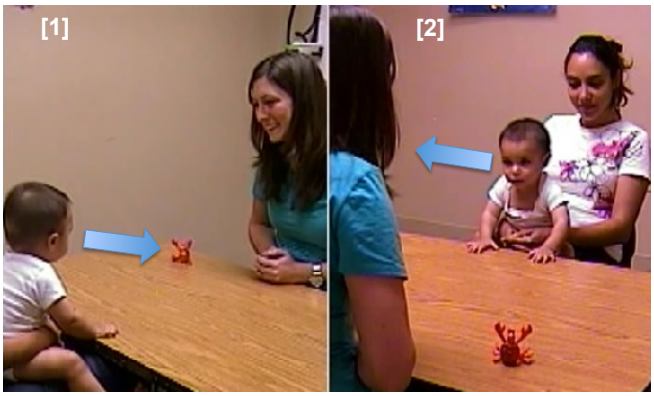
CONTACTO VISUAL PARA SEÑALAR UN INTERÉS

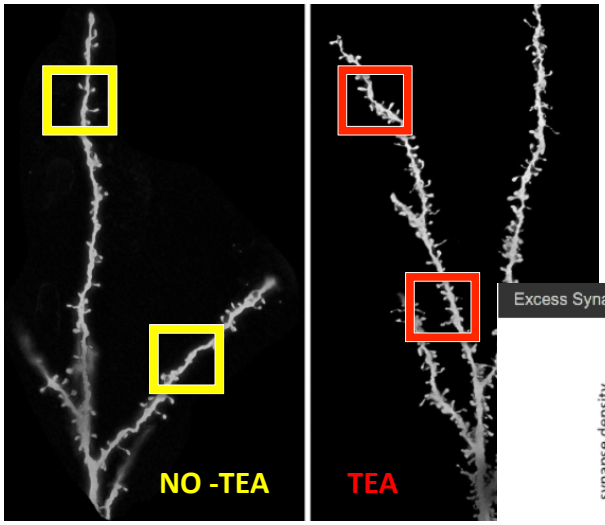


Sonrisa anticipatoria al objeto, seguida de sonrisa al adulto

LA DIFICULTAD PARA INTEGRAR LA SONRISA EN LA ATENCIÓN COMPARTIDA A LOS 8 MESES PREDICE LA SEVERIDAD DEL AUTISMO A LOS 30 MESES

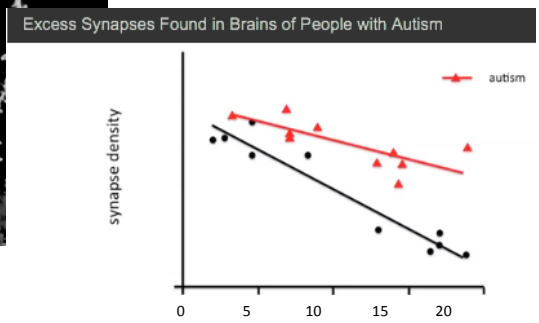
D. Gangi. JADD, June 2014

9



COLUMBIA UNIVERSITY MEDICAL CENTER

David Sulzer



"Loss of mTOR-dependent macroautophagy causes autistic-like synaptic pruning deficits."

G. Tang. *Neuron* Sep. 2014

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Gen RPTOR (Cr. 17)

Exceso de Actividad de Proteina **mTOR**

Disminución de la "poda" (50% vs. 16%) y "errores"

COLUMBIA UNIVERSITY MEDICAL CENTER

Restauración de la poda y mejora conductual

G. Tang. *Neuron* Sep. 2014

10

Problemas digestivos en niños y niñas con TEA

960 en total (TEA, DI y Controles)

TEA: 7 veces más de problemas digestivos

Dolor, gas, diarrea, estreñimiento y sensibilidad a alimentos

↓

+ irritación, + aislamiento, + problemas de conducta

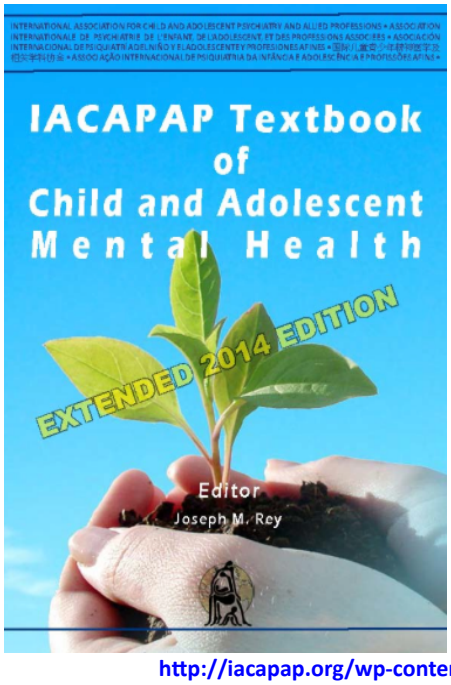
↓

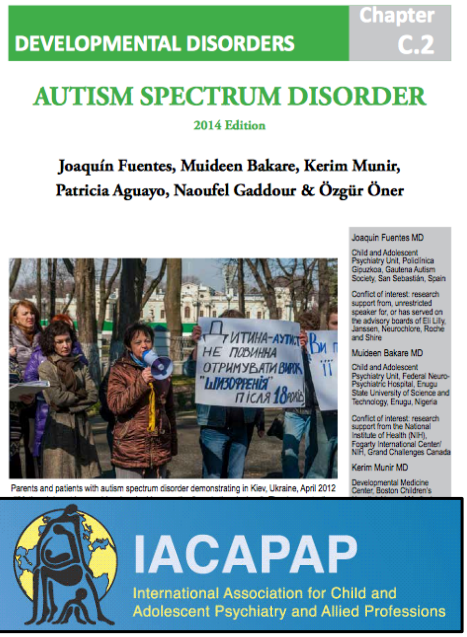
¿Hábitos y alimentación restrictiva?

↓

¿Molestias y ansiedad reactivas?

V. Chaidez. *JADD,* Nov 2013





http://iacapap.org/wp-content/uploads/C.2-AUTISM-SPECTRUM-072012.pdf

INSAR International Society for Autism Research

Start | Browse by Day | Browse by Format | Author Index | Disclosure Index

Note: Most Internet Explorer 8 users encounter issues playing the presentation videos. Please update your browser or use a different one if available.

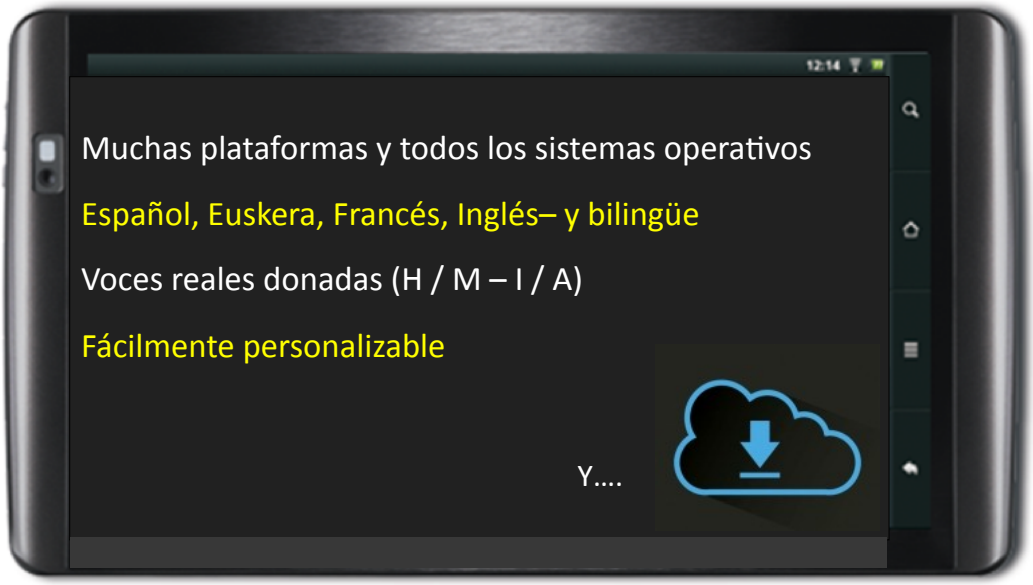
Preconference

Wednesday, 1 May 2013

Wednesday | top

12:00-13:00		
2755	The New Definition Of ASD (DSM5)	A. Martin
13:00-14:00		
2754	Understanding The Person With ASD	F. Happé
14:00-15:00		
2753	Explaining ASD: Genetic Aspects	T. Bourgeron
15:00-16:00		
2752	Early Diagnosis and Course in ASD	R. Landa
16:00-17:00		
2751	Explaining ASD: Neurobiological Aspects	J. Piven
17:00-18:00		
2750	Treatment Of ASD In Early Childhood	C. Kasari
18:00-19:00		
2749	Treatment Of ASD During The Life Cycle	P. Howlin
19:00-20:00		
2748	ASD: Support and Community Development	J. Fuentes

Hace cuatro años….



> 70.000 descargas desde 115 países

(Junio / 2015)

www.fundacionorange.es

http://www.policlinicagipuzkoa.com/e-mintza.php

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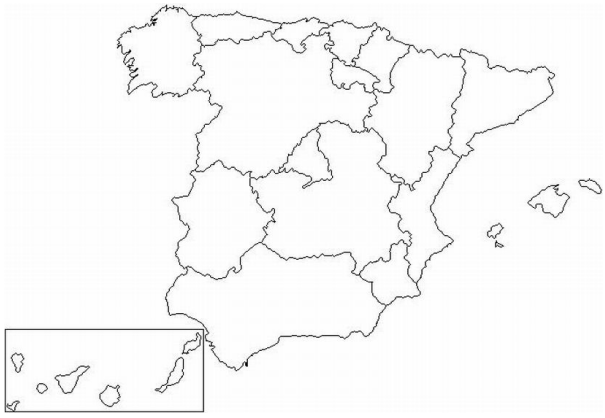
RETEA



REGISTRO DE LOS TRASTORNOS DEL ESPECTRO DEL AUTISMO

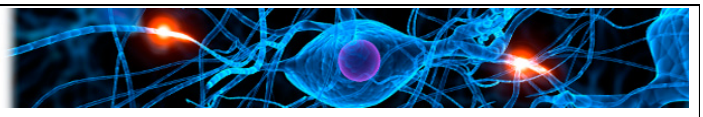
Registro de TEA (RETEA)

Declarado en la Agencia Española de Protección de Datos y como tal está sujeto a la normativa de la Ley Orgánica 15/1999, de 13 de diciembre, de Protección de Datos de Carácter Personal

Comité Científico

Manuel Posada de La Paz, IIER – ISCIII
Ignacio Abaitua Borda, IIER – ISCIII
Ricardo Canal Bedia, Universidad de Salamanca
Joaquín Fuentes Biggi, Policlínica Gipuzkoa y Gautena

EU-AIMS
Autism Research in Europe

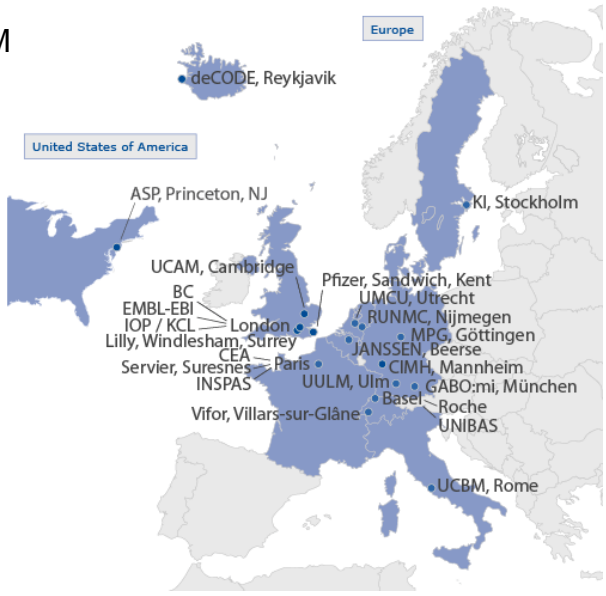


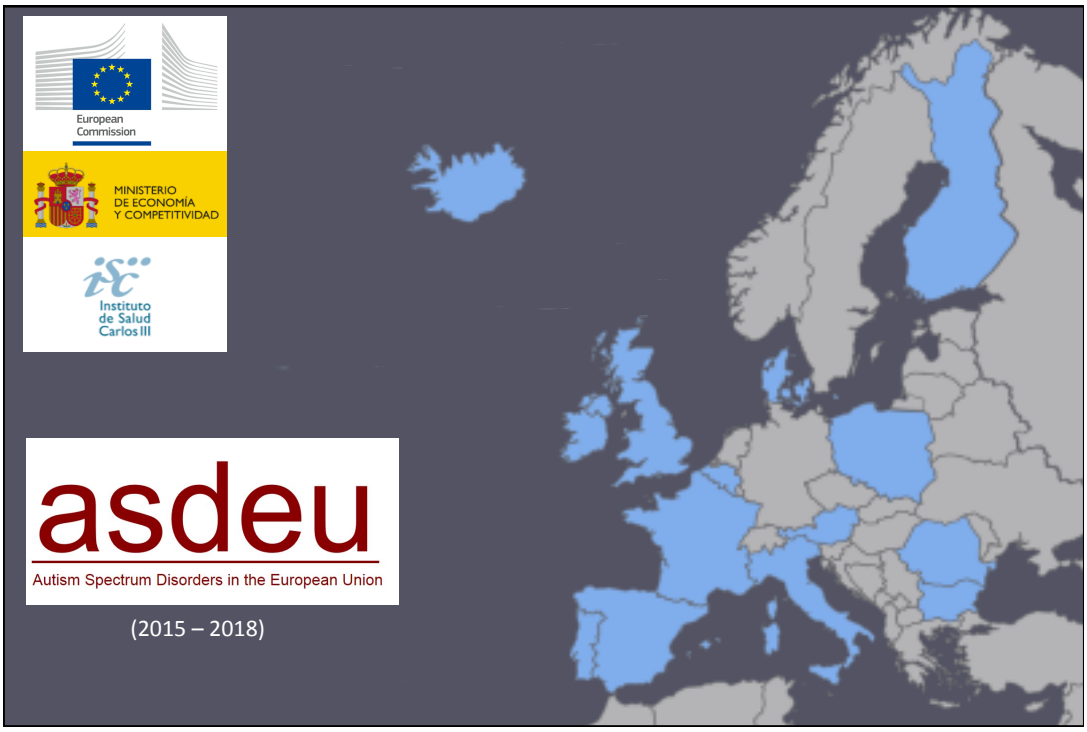
"… Para el final del 2017 esperamos ofrecer nuevos ensayos celulares validados, modelos animales, nuevas técnicas de resonancias magnéticas funcionales, nuevos radioligandos para la TEP, así como nuevos biomarcadores genéticos y proteómicos para la separación y agrupamiento de pacientes o para predecir la respuesta individualizada. Ofreceremos una red de investigación que pueda rápidamente testar nuevos tratamientos en seres humanos. Estas herramientas darán a los miembros de EFPIA una ventaja en el desarrollo de nuevos medicamentos para los TEA…"

30M€

EU-AIMS
Autism Research in Europe

CONSORTIUM




European Commission
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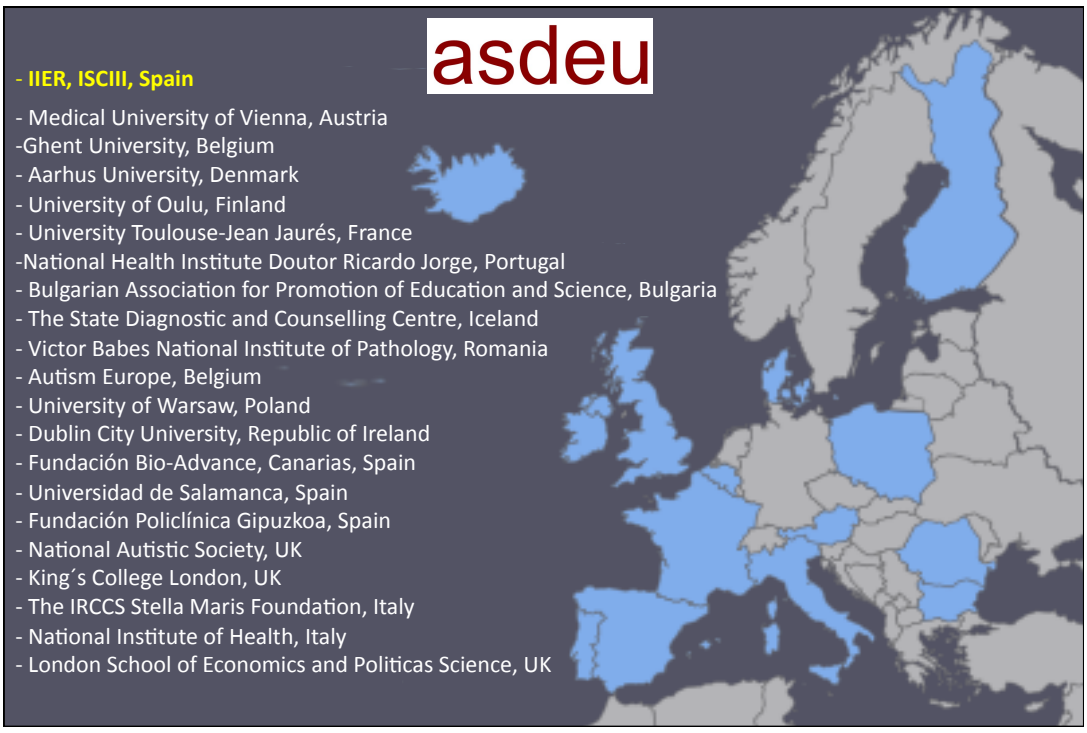
asdeu
Autism Spectrum Disorders in the European Union
(2015 – 2018)


asdeu
- IIER, ISCIII, Spain
- Medical University of Vienna, Austria
-Ghent University, Belgium
- Aarhus University, Denmark
- University of Oulu, Finland
- University Toulouse-Jean Jaurés, France
-National Health Institute Doutor Ricardo Jorge, Portugal
- Bulgarian Association for Promotion of Education and Science, Bulgaria
- The State Diagnostic and Counselling Centre, Iceland
- Victor Babes National Institute of Pathology, Romania
- Autism Europe, Belgium
- University of Warsaw, Poland
- Dublin City University, Republic of Ireland
- Fundación Bio-Advance, Canarias, Spain
- Universidad de Salamanca, Spain
- Fundación Policlínica Gipuzkoa, Spain
- National Autistic Society, UK
- King´s College London, UK
- The IRCCS Stella Maris Foundation, Italy
- National Institute of Health, Italy
- London School of Economics and Politicas Science, UK

European Commission

asdeu

Autism Spectrum Disorders in the European Union

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1. Prevalencia / Costes social y económico
2. Detección temprana, diagnóstico e intervención
3. Vida adulta con TEA – tratamientos y apoyo
4. Políticas para TEA en Europa – Salud Pública
5. Información y evaluación

J Am Acad Child Adolesc Psychiatry. 2014; 53:1145-1146

CLINICAL PERSPECTIVES

Autism Spectrum Disorders: Ten Tips to Support Me

Joaquin Fuentes, MD

The recently revised American Academy of Child and Adolescent Psychiatry Practice Parameter for the Assessment and Treatment of Children and Adolescents With Autism Spectrum Disorder[1] highlights the importance of clinicians maintaining an active role in family and individual support. Its evidence-based recommendations coincide with those of the International Association for Child and Adolescent Psychiatry and Allied Professions, the European Society for Child and Adolescent Psychiatry, and Autism Europe.[2] In contrast, in Europe, there is a greater emphasis on an approach to children and adolescents with autism spectrum disorder that is based on rights, participation, and quality. Inclusion Europe[3] leads a campaign for making information easily understandable as an essential mechanism to foster citizen participation, ensure informed choice, and protect human rights.

Recognizing the complementary strengths in these approaches, my colleagues and I have produced a tool to empower stakeholders, guide caregivers, and provide a rationale for advocates. The document was originally produced by its author and then reviewed, edited, and formally endorsed by a self-support group of young persons with Asperger disorder and by the Board of Families from the Cipuzkoa Autistic Society, the largest autism community program in southern Europe.

It is hoped that this document, also accessible in Basque, French, and Spanish in Supplement 1 (available online), will become a framework for clinical practice and global advocacy.

AUTISM SPECTRUM DISORDERS: 10 TIPS TO SUPPORT ME

1. I am not "autistic." I am first, foremost, and always a person, a student, a child, and I have autism. Do not confuse me with my condition. And, please, do not use the term in a negative or inconsiderate way. I deserve to be respected.
2. I am an individual. Having autism does not make me the same as other people with autism. Make an effort to know me as an individual, to understand my strengths, my weaknesses, and me. Ask me—and my friends and my family, if I cannot reply—about my dreams.
3. I deserve services, just like all children. Services for me begin early. Autism is—or it will be, when recognized—a public health issue in many countries of the world. There are instruments to screen it. They should be applied in the framework of screening for other developmental disabilities. If you start soon, my life will be different! And remember that about one quarter of my siblings will have autism or other problems. Help them; they are an important part of my life.
4. I belong in the health care system, just like all children. Include me in regular health care. The health care system should adapt to me, limiting waiting times and ensuring that I understand what is to be done, by using, for example, easy-to-read materials, pictograms, technologic means, and so forth. Other patients also will benefit.
5. I belong with other children. Do not separate me from them because you want to treat me, educate me, or care for me. I can, and I should, be placed in regular schools and regular community settings, and special support should be provided to me in those places. I have something to teach other children and something to learn from them.
6. I belong with my family. Plan with me for my future and my transitions. I am the one who should decide, and, when my ability to do so is limited, my family and friends will speak for me. No government agency can take the

Supplemental material cited in this article is available online.