On the *ADNP* gene and its clinical presentation

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nature genetics

A SWI/SNF-related autism syndrome caused by *de novo* mutations in *ADNP*

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2014

10 children

Archival Report

Biological Psychiatry: Celebrating 50 Years

Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in *ADNP*

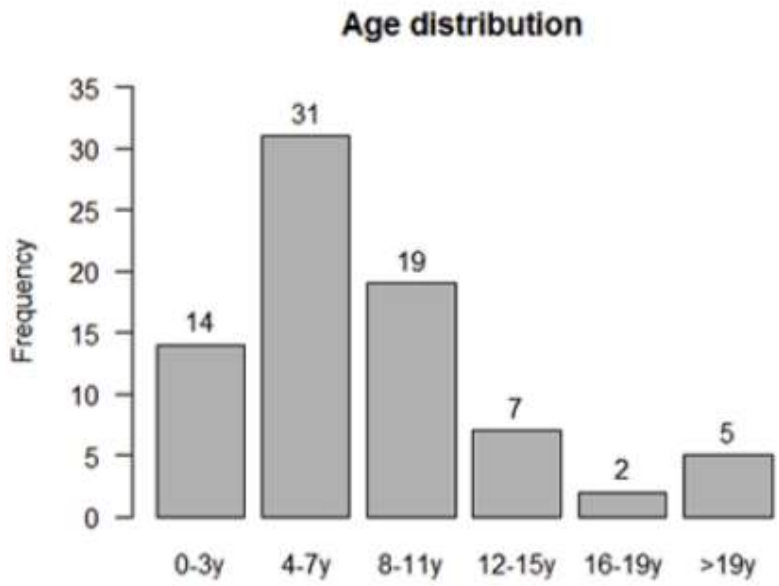
Anke Van Dijck, Anneke T. Vulto-van Silfhout, Elisa Cappuyns, Ilse M. van der Werf, Grazia M. Mancini, Andreas Tzschach, Raphael Bernier, Illana Gozes, Evan E. Eichler, Corrado Romano, Anna Lindstrand, Ann Nordgren, *ADNP* Consortium, Malin Kvarnung, Tjitske Kleefstra, Bert B.A. de Vries, Sébastien Küry, Jill A. Rosenfeld, Marije E. Meuwissen, Geert Vandeweyer, and R. Frank Kooy

2018

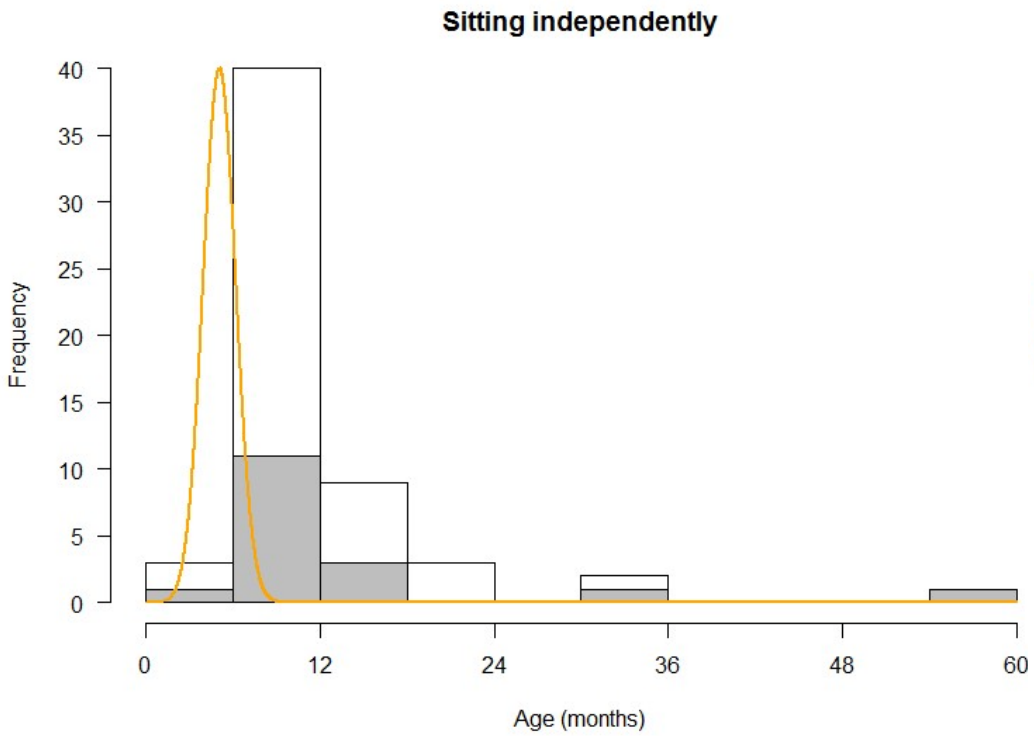
78 participants

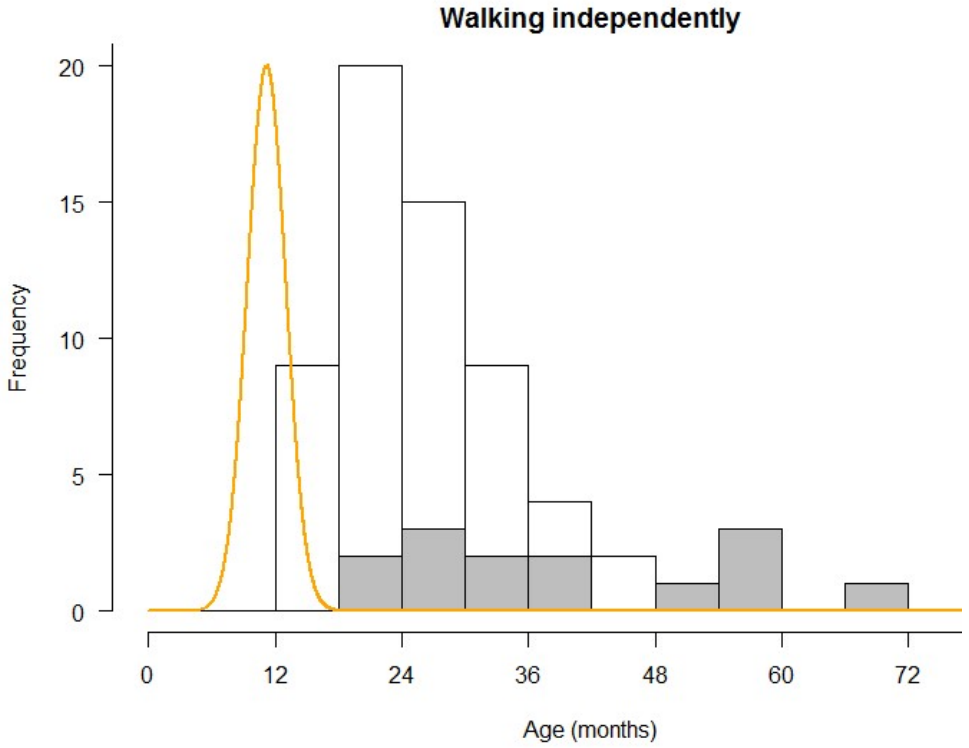
Clinical features

- Mild to severe intellectual disability
- Autism
- Severe speech and motor delay
- Characteristic facial features

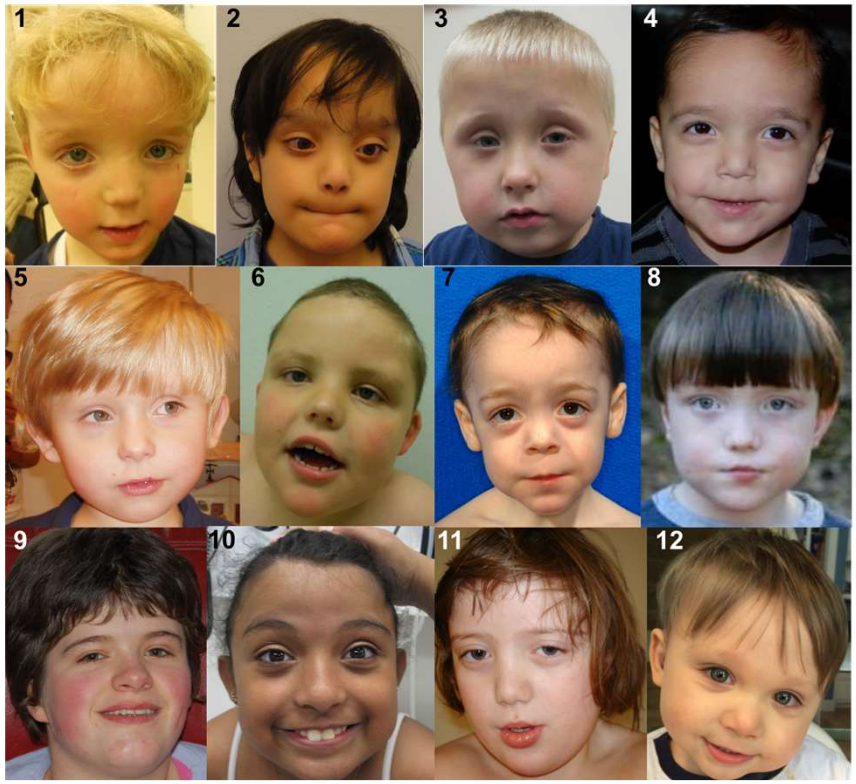


Development

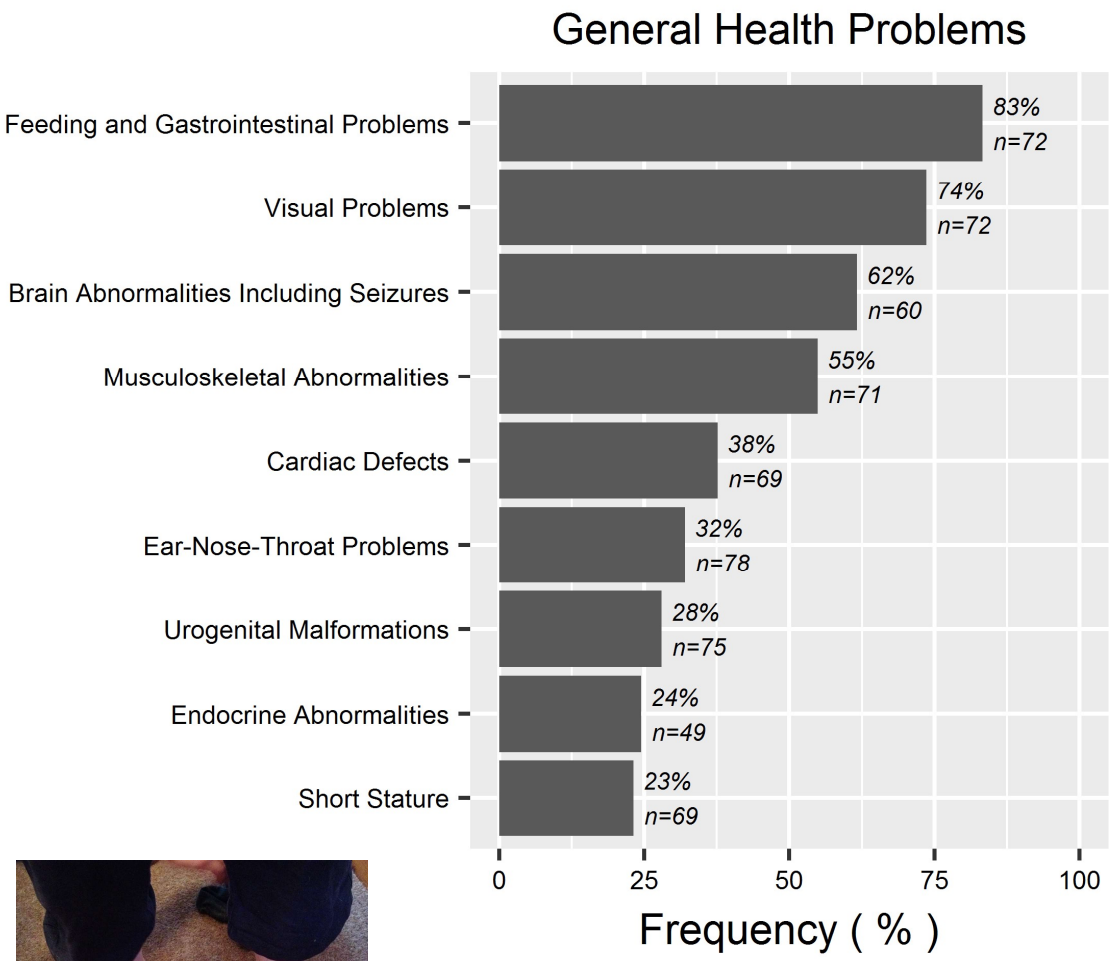


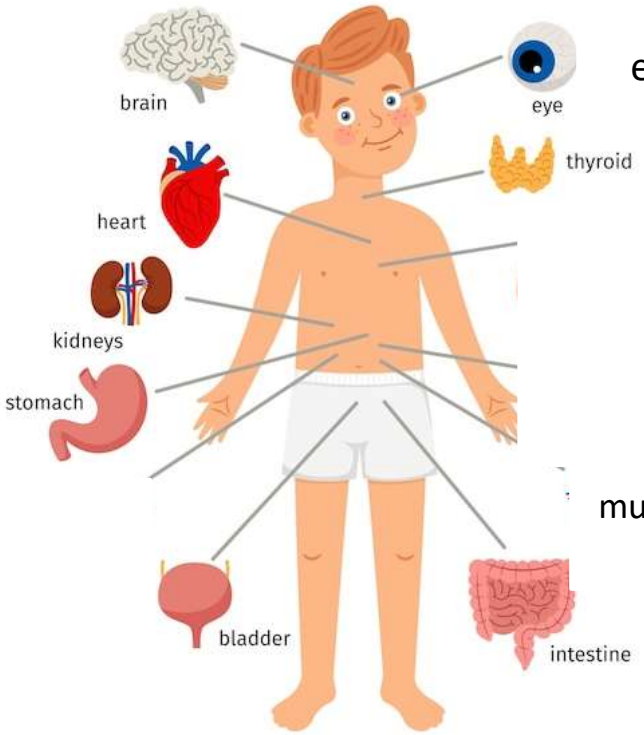


Facial features



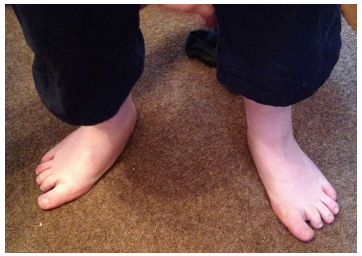
- Prominent forehead
- High anterior hairline
- Wide and depressed nasal bridge
- Short nose with full tip
- Thin upper lip, everted lower lip, pointed chin
- Ear malformations

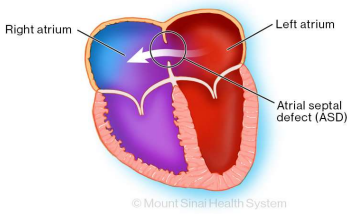


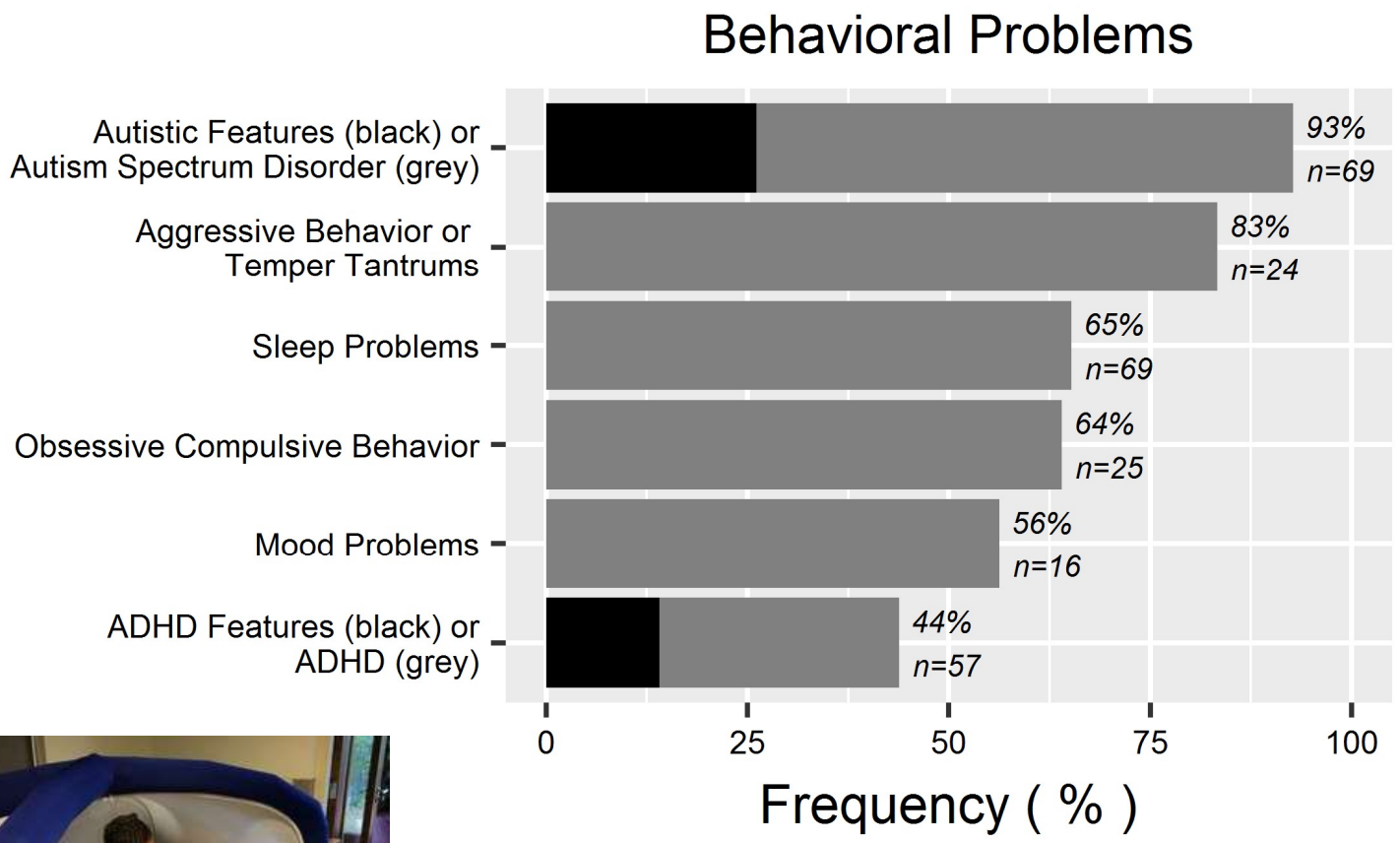


ears

muscles






Behavioral Problems
Autistic Features (black) or Autism Spectrum Disorder (grey)
Aggressive Behavior or Temper Tantrums
Sleep Problems
Obsessive Compulsive Behavior
Mood Problems
ADHD Features (black) or ADHD (grey)
93%
n=69
83%
n=24
65%
n=69
64%
n=25
56%
n=16
44%
n=57
0
25
50
75
100
Frequency (%)

Recommended evaluations/surveillance

Clinical geneticist
Pediatrician (+ blood test, including thyroid function)
Neurologist (+ if necessary EEG, brain MRI)
Cardiologist (+ Sonography, ECG)
Ophtalmologist (+ CVI investigation)
Physiotherapist
Speech therapist
Dietician
Psychologist / Psychiatrist

The International
ADNP Family Day

Sunday November, 6th 2016
Westcord Fashion Hotel Lounge Amsterdam,
The Netherlands

Thank you!

Literature

- Helsmoortel C, …, Kooy RF, Eichler EE, Van der Aa N. (2014). **A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP.** Nat Genet Apr;46(4):380-4.

- Van Dijck A, …, Kooy RF. **Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP**. Biol Psychiatry. 2019 Feb; 85(4) 287-297

Please ask your physician to submit clinical information on the website!

- https://humandiseasegenes.nl/adnp