ARTYKUŁ REDAKCYJNY
EDITORIAL

Medycyna Środowiskowa - Environmental Medicine 2016, Vol. 19, No. 1, 7-8
www.medycynasrodowiskowa.pl www.journal-em.com

Autism Spectrum Diseases: Genetics or Environment? Facts and Legends. Short look at the problem

Choroby ze spektrum autyzmu: genetyczne czy środowiskowe? Fakty i legendy. Rzut oka na problem

Karl Ernst v. Mühlendahl

Kinderumwelt gemeinnützige GmbH der Deutschen Akademie für Kinder- und Jugendmedizin (DAKJ) e.V., Osnabrück, Germany

Prof. Karl Ernst v. Mühlendahl

Measles-mumps vaccinations might be responsible for the development of autism spectrum diseases (ASD), including high functioning autism, such as Asperger's syndrome; that is what Wakefield et al. suggested in a paper in The Lancet twenty years ago [1]. Later, the Lancet Editors retracted this publication [2], due to the fact that Wakefield had „produced" fake, falsified results, and that he was implicated in interests regarding material compensation from pharmaceutical companies producing vaccines to families with ASD. The Wakefield publication had the effect that consequently vaccination rates declined dramatically, and fuelled discussions about other environmental factors (bacteria, mercury, pesticides, etc.).

GENETICS

Genetic factors are of paramount importance for the development of ASD. Analyses of more than thousand families with several members with ASD, and of a large number of families with one case only [3], have made the following observations apparent:

The incidence in boys or men is approximately three times as high as in girls or women.

If there are further children in families with two or more ASD cases, about 50% of sons, but only 20% of daughters will have ASD.

In monozygotic twins, there is a practically complete (96%) concordance in males, but in only 85% both girls have ASD.

In dizygotic twins, concordance is lower: 60% in males, 20% in females.

The marked difference between male and female gender points to a higher susceptibility in boys and/or to a postulated female protective factor (FPE) [4]

Key words: autism, children, environment

ENVIRONMENT

As genetically identical twins are relatively rare – this is the case namely in girls – and also for other reasons derived from genetic data, other factors – i.e. the environment – necessarily must play a role. They might alter chromosomal architecture (genes, DNS), and/or they could modify epigenetic factors which activate or suppress any existing genetic predisposition [5].

Such environmental factors most probably affect

Nadesłano: 5.02.2016
Zatwierdzono do druku: 22.02.2016

only genetically susceptible individuals in embryonal and fetal live or postnatally. Numerous conditions and potential toxicants have been considered to be the cause (table 1). Is it via modification of receptor activities, due to oxidative stress, or due to interference in hormonal mechanisms? Do testosterone and other androgens play a role, or is it because of environmental agents acting as endocrine disruptors? A recent large twin study discards the testosterone hypothesis [6].

Table I. Some alleged environmental causes for ASD, acting pre- or postnatally

- analgetics
- diabetes
- environmental "toxicants"
- infections
- mercury
- nutritional factors
- parental age
- perinatal stress
- pesticides
- prenatal stress
- proximity to highways
- testosterone, androgens
- vaccinations
- valproat
- zink deficiency

Functional magnetic nuclear resonance (NMR) investigations have shown, under certain experimental conditions, that there are activations in some cerebral areals in ASD persons, but not in controls [7].

There exist also speculations on the role of mirror neurons, a postulated cellular and functional system responsible for intuitive understanding, for pity and empathy. It is understandable that mirror neurons are discussed just in this context, but a recent review [8] discards these considerations.

In summary, there does not exist any valid, scientifically founded proof for any of the hypotheses regarding the role of environmental agents in the genesis of ASD.

FAIRY TALES

What makes a good fairy tale? It should be brief, it should have one single strain of action and one-dimensional reasoning; and it should fit into the listener's or reader's pattern of thinking. Wakefield's story is of such stuff, and this may be the reason for which it is retold over and over again, even today, despite its fraudulent origin.

REFERENCES

[1] Wakefield A.J. et al.: Ileal-lymphoid-nodular hyperplasia, non-specific colitis, and pervasive developmental disorder in children. Lancet 1998; 351: 637-641.
[2] Horton R.A.: A statement by the editors of The Lancet. Lancet 2004; 363: 820-821.
[3] Werling D.M., Geschwind D.H.: Recurrence rates provide evidence for sex-differential, familial genetic liability for autism spectrum disorders in multiplex families and twins. Mol Autism 2015; 6: 27-41.
[4] Mandy W., Lai M.C.: Annual research review: The role of the environment in the developmental psychopathology of autism spectrum condition. J Child Psychol Psychiatry; Jama.doi.10.1111/jcpp.1250; 2016.
[5] Koufaris C., Sismani C.: Modulation of the genome and epigenome of individuals susceptible to autism by environmental factors. Int J Mol Sci 2015; 16: 8699-8718.
[6] Eriksson J.M., Lundström S.: Lichtenstein P, Bejerot S, Eriksson E. Effect of co-twin gender on neurodevelopmental symptoms: a twin register study. Mol Autism 2016; 7: 8-13.
[7] Sun-Young K. et al.: Abnormal activation of the social brain network in children with autism spectrum disorder: An fMRI study. Psychiatry Investig 2015; 12: 37-45.
[8] Hamilton A.: Reflecting on the mirror neuron system in autism: A systematic review of current theories. Dev Cogn Neurosci 2013; 3: 91-105.

Address for correspondence:
Karl Ernst v. Mühlendahl
Kinderumwelt gemeinnützige GmbH der Deutschen Akademie für Kinder – und Jugendmedizin (DAKJ) e.V., Westerbreite 7, D 49084 Osnabrück, Germany
e-mail: info@uminfo.de