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Heritability and Genetic Relationship of Adult Self-Reported Stuttering, Cluttering and Childhood Speech-Language Disorders

Dziedziczność i genetyczne zależności jąkania, giełkotu i zaburzeń rozwoju języka w opisie osób dorosłych

SUMMARY

Genetic influence and mutual genetic relationship for adult self-reported childhood speech-language disorders, stuttering, and cluttering were studied. Using nationwide questionnaire answers from 34,944 adult Danish twins, a multivariate biometric analysis based on the liability-threshold model was performed in order to estimate heritability of the traits and genetic correlation between them.

The lifetime prevalence rates were in agreement with previous reports, and were higher for males than for females for all three traits. The probandwise concordance rates were always substantially higher for monozygotic compared to dizygotic pairs, suggesting genetic influence. Multivariate biometric analyses showed that additive genetic and unique environmental factors best explained the observed concordance patterns. Heritability estimates for males/females were 0.71/0.87 for childhood speech-language disorders, 0.78/0.80 for stuttering, and 0.53/0.65 for cluttering. For each trait, the same genes were suggested to affect liability in males and females. Furthermore, high genetic correlations between the traits were obtained; the estimates for childhood speech-language disorders and stuttering were 0.71/0.79 for males/females, for childhood speech-language disorders and cluttering 0.73/0.56, and for stuttering and cluttering 0.53/0.57.

Substantial unique environmental correlations between the traits were also found in both genders.

Conclusion: With the limitations related to self-reporting from adult age, this study demonstrates substantial genetic influence on the traits of childhood speech-language disorders, stuttering, and cluttering, and mutual genetic relationship between them.

INTRODUCTION

We aimed to investigate the mutual relationship between stuttering, cluttering and childhood speech-language disorders at the genetic level

by asking single item questions. Our intension is to validate if this simple one-item questions agree with results from screened or diagnosed entry data. If the agreement is found we like to expand the biometric model to a new aspect of the relation between the three traits of developmental oral communication disorders.

MUTUAL RELATIONSHIP BETWEEN, STUTTERING, CLUTTERING AND CHILDHOOD SPEECH-LANGUAGE DISORDERS

Simultaneously occurrence of stuttering and childhood speech-language disorders, stuttering and cluttering as well as cluttering and childhood speech-language disorders has been claimed and described through clinical communications by Treitel (1892), Gutzmann (1893), Liebmann (1900), Scripture (1912), Weiss (1935), Pichon & Borel-Maisonny (1937), De Hirsch and Langford (1950), Gedda, Bracconi & Bruno (1960), Luchsinger (1963), and Van Riper (1971). Nadoleczny (1929) expressed the view that stuttering is frequently based on a hereditary weakness of the disposition to speech, but Watkins (2005) describes that groups of young children who stutter display expressive language abilities at or above normative expectations. Freund (1952) noted that both stuttering and cluttering often appeared simultaneously in family members and hypothesized that a hereditary element is involved, so did Weiss (1950). Weiss (1964) also mentions the comorbidity between cluttering and delayed speech development. Both stuttering and cluttering have been regarded as disorders of speech motor control (Kent, 2000) and language factors may be important in both disorders (Kent, 2000; Guitar, 2006, 61). The two disorders affect more males than females, and possibly gender-related differences in brain maturation account at least in part for the preponderance of males in populations with developmental apraxia of speech (and stuttering as well) (Kent, 2000).

St. Louis, Myers, Bakker & Raphael (2007) refer a general agreement, that cluttering often co-exists with other fluency disorders, rate deviations, stuttering, articulation disorders, language disorders, Attention Deficit Hyperactivity Disorders (ADHD), specific learning disabilities, Central Auditory Processing Disorders (CAPD), basal ganglia syndrome and/or speech apraxia, although St. Louis, Raphael, Myers & Bakker (2003) mentioned the confusing issue that cluttering often – but not always – coexists with stuttering. Therefore St. Louis et al. (2003) concluded that the two fluency disorders are now regarded as distinct fluency disorders by most authorities. Furthermore, the available literature on differentiating cluttering and stuttering suggests that the essential difference between

these clinical populations centers on the speaker's level of preparedness for saying intended utterances. Stutterers know what they want to say but are interfered in their attempt to produce various words, whereas clutterers do not necessarily know all what they want to say – or how – but say it anyway (St. Louis, et al, 2007).

On this background we might find some kind of shared genetic or environmental pathogenesis for the three traits. To investigate the possible mutual relationship between the self-evaluated traits of stuttering, cluttering and childhood speech-language disorders we use the twin method. Our approach is to simultaneously estimate, by multivariate modelling, the heritability of each of the traits and the genetic correlation between them.

CHILDHOOD SPEECH-LANGUAGE DISORDERS

Seeman (1937) was the first to be convinced that genetic factors often are responsible for various delays in language development, and Ingram (1959) noticed genetic determined heredity of specific developmental disorders of speech in childhood. Arnold (1961) described three families with a large number of children with disorders of language learning. Heritability estimates for specific language impairment have been inconsistent, with monozygotic (MZ) probandwise concordance rates between 0.36 and 0.96, and dizygotic (DZ) rates between 0.20 and 0.69. Genetic studies are more likely to find high heritability if they focus on cases who have speech difficulties and who have been referred for intervention (Bishop & Hayiou-Thomas, 2008). Bivariate genetic analysis estimated a genetic correlation of 0.63 between general language and nonverbal factors, implying that over half of the genetic influence on language overlaps with genetic influence on nonverbal factors (Colledge, 2002). On the other hand only modest heritability was found for individual differences in the normal range in 4-year-old same-sex and opposite-sex twins (Viding et al., 2004). Spinath et al. (2004) found MZ similarity intraclass correlations for children's early language ability were greater than DZ correlations suggesting genetic influence. Genetic influences were numerically greater for boys in all analyses. Probandwise twin concordance for low language ability shows a clear pattern with substantially greater MZ twin concordance compared with same gender DZ twins regardless of gender, indicating genetic influences on the risk that cotwins of probands are themselves affected. For the aggregated measure, a weighted average of the probandwise twin concordance yielded 0.86 for MZ twins and 0.52 for same gender DZ twins. So, there is a 86% risk that the cotwin is also low in language ability.

For DZ twins, the risk reduces to 52%. In general the gender effects were small, but significant, and there was an indication that the role of genes is stronger for boys than for girls.

In the Danish language childhood speech problems (taleproblemer som barn) is used for the English expression speech and language delay and disorders in childhood, and the Danish expression for language problems (sprogproblemer) refer to problems or difficulties with a second language. This is documented in the in *Korpus 2000,* a project to document the use of Danish language between 1998 and 2002 (http://korpus.dsl.dk/korpus2000/engelsk_hovedside.php3?lang=dkaround, Andersen, Asmussen & Asmussen, 2002). In our study, based on self reported questionnaire, we therefore use the term childhood speech problems (taleproblemer som barn), which in a Danish context cover the term speech and language delay and disorders.

STUTTERING

The genetic contributions to stuttering have long been highlighted by classic genetic studies; see Yairi, Ambrose & Cox (1996) and Yairi & Ambrose (2005) for critical reviews. Twin studies have been relatively few in number and with small sample sizes, and older studies on stuttering in twins seem to suggest that there are relatively more stutterers among twins than in the general population. However, there is a lack of agreement on the difference and other aspects. Berry (1937 & 1938) reported that stuttering is more frequent among the twins (one out of 11) than the singletons (one out of 35) in twinning families, and a correlation trend between lefthandness, twinning and stuttering. Nelson, Hunter & Walter (1945) reported stuttering in 20 per cent of 200 twin pairs, and a lower male to female stuttering rate in twins than singletons. They also found that concordant pairs with stuttering were more likely in monozygotic than dizygotic twins, and so did Seeman (1937) and Luchsinger (1940) with few cases, but it was also confirmed by Graf (1955). Berry (1937) reported greater incidence of twins in families containing stutterers, and Wepman (1939) reported more stuttering in families having twins. Graf (1955) studied 552 pairs of twins from a population of 85,680 pupils from the public schools and found that 1.90 per cent of the twins stuttered, and in seven out of ten pairs with one stuttering individual, also twin B stuttered. The classic genetic analysis was performed by Howie (1981) who showed higher pairwise and probandwise concordance rates in MZ pairs in 30 same-gender twin pairs found in public speech clinic files and

in response to a newspaper request. In general, the relatively higher rate of stuttering among twins than singletons has historically been explained with reference to hereditary factors.

Some population based studies on twins and stuttering have been published and genetic factors have been established for stuttering with higher concordances for monozygotic (MZ) compared with dizygotic (DZ) twin pairs using unselected community-based adult twin samples (Andrews et al., 1991 & Felsenfeld et. al., 2000); Andrews et al. with 71% of the variance attributed to additive genetic variance.

In the study of Felsenfeld et al. a large population-based twin sample from the Australian Twin Registry (1567 pairs and 634 singles aged 17–29 years) was screened to identify twin pairs in which one or both members reported themselves to be affected by stuttering. Telephone interview-based diagnoses were obtained for 457 of these individuals (self-reported affected cases, cotwins, and controls) to determine whether the self-report was correct. To correct for ascertainment bias they carried out a bivariate analysis of the final diagnosis in the selected sample with the screening item in the full sample, using maximum-likelihood methods for raw ordinal data implemented in Mx 1.47c (Neale et al., 2006). After correcting for ascertainment bias, approximately 70% (95% confidence interval: 39–86%) of the variance in liability to stuttering was found to be attributable to additive genetic effects, with the remainder 30% due to non-shared environmental effects.

Oliver & Plomin (2007) found consistent and moderate genetic and non-shared environmental influences, and modest common environmental influence. Dworzynski (2007) concluded that stuttering appears to be a disorder that has high heritability and little common environment effect in early childhood, and also for recovered and persistent groups of children by age 7. Using questionnaire data Ooki (2005) reported that total phenotypic variance attributable to heritability was 80% for boys and 85% for girls at an average age of 11.6 years. Probandwise concordance rates were 0.52 for MZ and 0.12 for same gender DZ, and polychoric correlations were 0.81 for MZ and 0.18 for DZ (all values slightly higher for females than males). However, concordances themselves cannot be used to estimate genetic and environmental parameters because they do not take into account population prevalence rates (Dworzynski, 2007). Yari and Ambrose (2005) have summarized the contribution of genetics to stuttering and the best-fitting transmission model for stuttering. Both genetic and environmental factors contribute to stuttering.

NO SINGLE STUTTERING SUSCEPTIBILITY LOCUS

Stuttering is regarded as a multifactorial-polygenic disorder with many contributing loci of varying effects, and gene by environment interaction. LOD score of 2.20 has been achieved on chromosome 7 at D7S559 (181.97 cM) on the most telomeric marker on the chromosome arm (Riaz, 2005) – but final markers give less precisely evidence for linkage. Chromosome 7 has shown nominal evidence for linkage with LOD score of 1.69, and for male-only data 2.99 at 153 cM (P = .04) (Suresh et al., 2006). Shugart et al. (2004) suggested that chromosome 18 may harbour a predisposing locus for stuttering and that stuttering may display locus heterogeneity in different study populations. Meta-analysis for stuttering has identified 12 broad regions on chromosomes 2, 3, 5, 7, 9, 13 and 15 showing nominal significant evidence for linkage to stuttering (Wittke-Thompson et al. 2007). Recently, Kang et al. (2010) identified in persons who stuttered a missense mutation in the 12q23.3 genomic region in 10% of consanguineous Pakistani families with stuttering members.

CLUTTERING

Cluttering is not so well described as stuttering and childhood speech-language disorders, and Colombat was sited for reporting the first differentiation between cluttering and stuttering in 1830, according to St. Louis, Hinzman & Hull (1985). Still, the nosologic status of cluttering is open to much debate (Kent, 2000), but Weiss (1964) and Arnold (1965) concluded that heredity plays a prominent role in most cases of cluttered speech. In comparison with other types of disturbed language function, cluttering is reported to occur about four times more often in males than in females at all ages. This gender difference also points to constitutional factors. Based on a large number of observations, Arnold suggested that two types of hereditary influences may be distinguished; specific and non-specific inheritance (Arnold 1958, 1960). Specific inheritance brings about the transmission of the cluttering syndrome in families containing many clutterers and stutterers, and Pfändler (1960) demonstrated an irregular dominant inheritance of cluttering. Non-specific inheritance manifests itself in the transmission of general language disability, including frequent occurrence of delayed speech, dyslalia, dysgrammatism, dyslexia or dysgraphia. Becker and Grundmann (1970) refer to a graduate thesis by Dietsch (1968) concerning heredity and cluttering. Dietsch interviewed the parents of 20 children suspected of cluttering at the age of seven or eight years who attended an ordinary school. The interviews had special attention

on the psychophysical development, the environmental and familiar situation, and heredity. In 17 of the 20 children hereditary factors were ascertained through an interview with the parents. In nine cases heredity was found to be specific, whereas for the remaining eight suspected of cluttering heredity was of an unspecific kind, such as weakness of speech disposition, nervousness, epilepsy, and equivalent phenomena.

Much focus has been on cluttering among speech-language pathologists in recent years but in the general public cluttering is not so well established as a disorder. In our study, based on self-reported questionnaire, we have tried to explain the trait we were asking for.

MATERIALS AND METHODS

The study is based on data from a large twin omnibus survey in 2002, reported by Fibiger, Tranebjærg & Skytthe (2004). This survey included also single item questions about stuttering, cluttering and childhood speech-language disorders, and the survey was organized by the Danish Twin Registry (DTR). The DTR is population based and was established 1954. It comprises more than 75,000 twin pairs born in Denmark since 1870. Almost all twins born are included, and ascertained independently of any diseases (Skytthe, et al., 2002; Skytthe, et al., 2006). The Registry contains information on health, diseases and causes of death.

SUBJECTS AND QUESTIONNAIRE

Based on the information given in the Introduction section, we investigated to what extent the co-existence of stuttering, cluttering and childhood speech-language disorders may result from a genetic vulnerability common to all three traits. To this end, we used the survey data mentioned above (Fibiger et al., 2004). A paper and pencil questionnaire was sent to a population-wide cohort of 46,418 twins, who had participated in earlier questionnaire studies and were born 1931 to 1982 in Denmark. The twins had been classified as monozygotic (MZ), same-gender dizygotic (SSDZ), or opposite-gender dizygotic (OSDZ) based on answers to four questions on physical similarity used for zygosity assessment in the Danish Twin Registry (Christiansen et al., 2003). The questionnaire was a 20 page A4 booklet with 119 main questions concerning: functioning, activity, disability, health, diseases, education, occupation, weight and length; tobacco smoking, alcohol consumption, family relations and children; fertility, thoughts and emotions (Skytthe, et al., 2006). Eleven research groups made this common questionnaire

and each research group could have access to all relevant answers from the common questionnaire. Some questions were general and not related to one specific research project. Twelve questions were used to report for disabilities and impairments related to hearing, speech & language, and reading including self-experience and self-reporting of otitis media, hearing problems, hearing aid, Ménière's disease, tinnitus, cryptophasia, childhood speech-language disorders, stuttering, cluttering, acquired speech disorders, aphasia, and dyslexia.

We used three questions to self-identify the twins for childhood speech-language disorders, stuttering and cluttering:

- „Did you have problems with your speech and language in your childhood?"
- „Do you stutter or have you stuttered?"
- „Is it, or has it been a problem, that you speak so fast, that you stumble over the words and omit syllables (cluttering)?"

Speech-language pathologists differentiate between many sub-groups of speech and language problems and lack of development progression. Our intension in this study is to describe the heritability of the personally experienced communication problems. The drawback of self-evaluation is that it is far more subjective than psychometric assessments, but self-evaluation have the advantage that it enable us to obtain impressions from the person itself, and allow us to evaluate behaviours that may be difficult to elicit in a clinical setting.

Of the 46,418 questionnaires, 35,312 (76%) were returned by mail, with 33,794 twins being MZ, SSDZ or OSDZ. Of those twins, 32,548 (10,618 complete pairs, 11,312 unmatched twins) answered the question on childhood speech-language disorders, 33,317 (11,108 complete pairs, 11,101 unmatched twins) answered the question on stuttering, and 33,308 (11,084 complete pairs, 11,140 unmatched twins) answered the question on cluttering.

The answers to these three questions were self-reported, and behavior identification was based on self-experienced behavior and not a clinical diagnosis. There were no inclusion or exclusion criteria, but the majority of persons who have experienced childhood speech-language disorders, stuttering or cluttering has this behavior as a primary developmental disorder, not secondary to other developmental disorders (e.g. intellectual impairment, cerebral palsy). Possible comorbidity with other developmental disorders (e.g. ADHD) is not considered since this diagnosis was classified in DSM-III-R, 1987, and still only about 20 per cent of children with ADHD are diagnosed in Denmark. Childhood speech disorders have been treated in Denmark since 1898, so childhood speech-

language disorders are closely related to childhood, although many children with childhood speech-language disorders develop reading and writing problems. Stuttering and cluttering persist much more often also in adolescence and adulthood. Acquired stuttering and cluttering in adolescence and adulthood are extremely rare.

STATISTICAL ANALYSIS

Summary counts along with lifetime prevalence rates of each trait by gender and zygosity (MZ, SSDZ and OSDZ) were computed using the STATA software, version 9.

For each trait, probandwise concordance rates and tetrachoric correlations were estimated separately for MZ and SSDZ pairs, and for males and females.

Probandwise concordance is the probability that the trait occurs in a twin given that it has already occurred in the co-twin, and can be estimated as $2n_{11}/(2n_{11}+n_d)$, where n_{11} and n_d are the numbers of concordant and discordant twin pairs, respectively (Witte, Carlin & Hopper, 1999). Difference in concordance rate between MZ and DZ pairs suggests genetic effects.

Tetrachoric correlation is defined under the so called "liability-threshold" model. According to this model, there exists a latent liability to the trait, bivariate normally distributed in the population, with a threshold such that the trait occurs when the individual liability level exceeds the threshold. Tetrachoric correlation is the correlation in twin liabilities to the trait (Neale & Cardon, 1992), and is independent of trait prevalence. A significantly higher correlation in MZ compared to DZ pairs points to genetic influences on liability to the trait. To estimate tetrachoric correlations, saturated models were fitted to raw dichotomous data on each trait with the software Mx (Neale, Boker, Xie & Maes, 2006). These models were specified constraining the threshold of the trait to be the same for twin and co-twin, MZ and DZ twins.

MULTIVARIATE BIOMETRIC ANALYSIS

A multivariate sex-limitation Cholesky decomposition (Neale & Cardon, 1992), under the "liability-threshold" model, was applied to childhood speech-language disorders, stuttering, and cluttering, with the objective to estimate the genetic effects on each of the traits, and the degree of genetic overlap between them. In this model, not only the variance in liability to each trait but also the covariance between liabilities to two

different traits is decomposed into a sum of additive genetic (A), either non-additive genetic (D) or common (shared) environmental (C), and unique (unshared) environmental (E) components (fig. 1 – only A and E latent sources are displayed, for simplicity). Additive genetic influences originate from the additive effects of alleles at all contributing genetic loci, without allelic or gene-gene interaction. Dominance (allelic interaction within a gene) or epistasis (gene-gene interaction) are responsible for non-additive genetic effects. Shared environmental influences relate to exposures that are common to all members of a family. Unshared environmental factors are those factors that are specific to an individual, thus contributing to differences between family members; measurement error is also included in this latent source.

For each latent source (A, E), three independent factors are specified: the first (A_1, E_1) loads on all traits, the second (A_2, E_2) affects all traits except

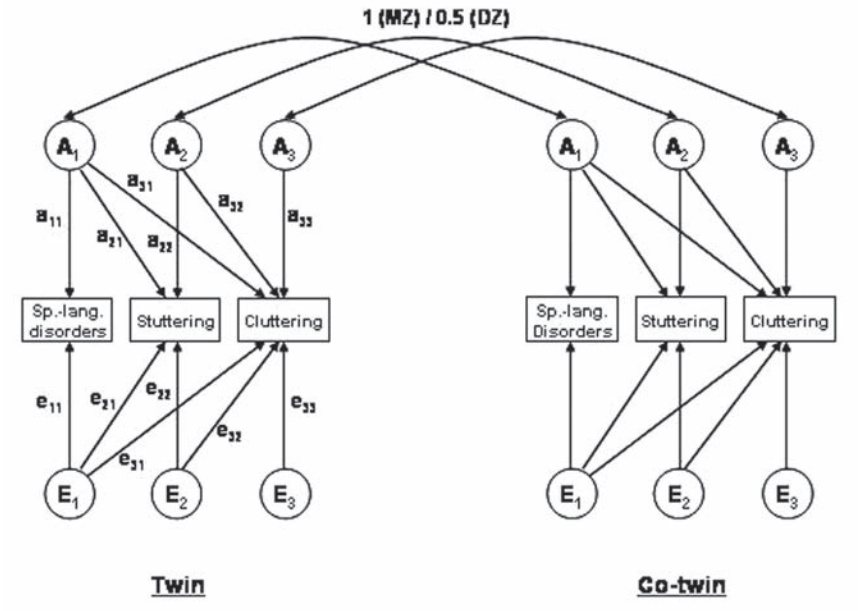

Fig. 1. Path diagram of the Cholesky decomposition for childhood speech-language disorders, stuttering, and cluttering in twin (left) and co-twin (right). Rectangles denote observed traits. Circles indicate latent sources of variance and covariance. A_i and E_i represent additive genetic and unique environmental influences on the traits. Reported beside the arrows are path coefficients. Additive genetic factors correlate 1 between MZ twins and 0.5 between DZ twins. Although model fitting also included non-additive genetic influences (D_i), the corresponding latent source was not shown in the diagram for reasons of clarity.

the first, the third (A_3, E_3) impacts on the last trait only; this formalizes the assumption that there may exist genetic and environmental influences common to the traits, supplemented by trait-specific effects.

This parameterization allows for the partition of the variances and covariances in liabilities to the traits into genetic and environmental components. For example, variances (Var) of childhood speech-language disorders and stuttering and their covariance (Cov) can be written as:

Var(childhood speech-language disorders) = $a_{11}^2 + e_{11}^2$
Var(stuttering) = $(a_{21}^2 + a_{22}^2) + (e_{21}^2 + e_{22}^2)$
Cov(childhood speech-language disorders, stuttering) = $a_{11}a_{21} + e_{11}e_{21}$ (Neale & Cardon, 1992).

Relevant statistics that can be derived from the above equations include: (i) heritability (h^2) of childhood speech-language disorders {[h^2(childhood speech-language disorders) = $a_{11}^2/(a_{11}^2 + e_{11}^2)$]} and of stuttering {$h^2$(stuttering) = $(a_{21}^2 + a_{22}^2)/[(a_{21}^2 + a_{22}^2) + (e_{21}^2 + e_{22}^2)]$}, defined as the proportion of variance due to genetic factors; (ii) genetic correlation {$r_A = a_{11}a_{21}/[a_{11}^2(a_{21}^2 + a_{22}^2)]^{1/2}$} and unique environmental correlation {$r_E = e_{11}e_{21}/[e_{11}^2(e_{21}^2 + e_{22}^2)]^{1/2}$} between childhood speech-language disorders and stuttering.

The quantity (i) is informative on the impact of inter-individual genetic differences in terms of inter-individual phenotypic differences in a single trait. The quantity (ii) can be regarded as a measure of the extent to which genes or environmental factors affecting liability to childhood speech-language disorders and to stuttering overlap, and thus gives information on the contribution of genes and environment to the co-morbidity of the two disorders. The overlapping between genes influencing different traits is known as genetic pleiotropy. For instance, if childhood speech-language disorders and stuttering are affected by independent sets of genes, the genetic correlation is zero. Evidence of pleiotropy is provided by a genetic correlation significantly different from zero.

Model fitting was performed using the Mx program (Neale et al., 2006). Due to the high computational burden, the models could not be fitted to raw dichotomous data via the maximum likelihood approach but were fitted to tetrachoric correlation matrices in MZ male, MZ female, DZ male, DZ female, DZ male-female, and DZ female-male twin pairs, using the asymptotic weighted least square method.

Model fitting started with a full ADE model, and then proceeded with a series of sub-models to test the significance of specific parameters by hierarchical chi^2 tests. The most parsimonious solution (best-fitting model) was used to derive model parameter estimates.

RESULTS

Out of 20878 MZ, SSDZ and OSDZ twins (10439 twin pairs) 1580 twins answered yes to childhood speech-language disorders, 1080 yes to stuttering, and 2361 yes to cluttering. 612 twins answered yes to childhood speech-language disorders and stuttering, 669 twins answered yes to childhood speech-language disorders and cluttering, and 483 twins answered yes to stuttering and cluttering. 319 twins answered yes to all three traits.

No significant differences emerged between twins from complete pairs and twins from unmatched pairs with respect to age, gender, zygosity, or lifetime prevalence rates for the disorders or phenotypic correlations for the traits.

Summary counts of twins who reported childhood speech-language disorders, stuttering and cluttering, and lifetime prevalence rates for childhood speech-language disorders, stuttering, and cluttering by zygosity and gender are given in table 1. Childhood speech-language disorders and stuttering were significantly more common in males than in females, while only a slightly higher prevalence in males was observed for cluttering. For stuttering, prevalences agreed well for MZ and DZ twins. Slightly higher prevalences for childhood speech-language disorders and cluttering in MZ twins compared to DZ twins were observed.

Table 2 shows, separately for MZ and SSDZ pairs and for males and females, the numbers of complete twin pairs and of concordant and discordant pairs, along with prevalence rates, probandwise concordance rates, and tetrachoric correlations for childhood speech-language disorders, stuttering, and cluttering.

Tetrachoric correlations were estimated under univariate saturated models specified with appropriate constraints (see the section 'Statistical Analysis'). Hierarchical chi^2 tests showed that the fit of these models was not significantly worse than that of the more general models without the constraints.

For all traits, both concordance rate and tetrachoric correlation were significantly higher in MZ than in SSDZ pairs, indicating substantial genetic influence on individual liability to each disorder. For stuttering, tetrachoric correlations suggested possible non-additive genetic effects.

Tab. 1. Summary counts and prevalence rates of childhood speech-language disorders, stuttering, and cluttering, by zygosity and gender

Trait	Zygosity*	Number of reported negative or positive answers to this question	Number of positive answers to this question	Lifetime prevalence**
		Males from same-gender pairs		
Speech-language disorders	MZ	3830	472	0.1232 (0.1131,0.1342)
Speech-language disorders	SSDZ	5854	565	0.0965 (0.0891,0.1044)
Stuttering	MZ	3936	325	0.0826 (0.0743,0.0917)
Stuttering	SSDZ	6005	498	0.0829 (0.0761,0.0903)
Cluttering	MZ	3934	595	0.1512 (0.1403,0.1629)
Cluttering	SSDZ	6010	756	0.1258 (0.1176,0.1345)
		Females from same-gender pairs		
Speech-language disorders	MZ	4849	325	0.0670 (0.0602,0.0745)
Speech-language disorders	SSDZ	6557	413	0.0630 (0.0573,0.0692)
Stuttering	MZ	4947	187	0.0378 (0.0327,0.0436)
Stuttering	SSDZ	6674	245	0.0367 (0.0324,0.0416)
Cluttering	MZ	4943	662	0.1339 (0.1246,0.1438)
Cluttering	SSDZ	6682	754	0.1128 (0.1054,0.1207)
		Males (M) and Females (F) from opposite-gender DZ pairs		
Speech-language disorders	OSDZ (M)	5060	507	0.1002 (0.0921,0.1089)
Speech-language disorders	OSDZ (F)	6398	277	0.0433 (0.0385,0.0486)
Stuttering	OSDZ (M)	5197	458	0.0881(0.9038,0.9194)
Stuttering	OSDZ (F)	6558	185	0.0282 (0.0244,0.0326)
Cluttering	OSDZ (M)	5197	458	0.0881 (0.0804,0.0958)
Cluttering	OSDZ (F)	6552	639	0.0975 (0.0905,0.1050)

*MZ = Monozygotic twins, SSDZ = Dizygotic twins from same-gender pairs, OSDZ = Dizygotic twins from opposite-gender pairs.
**In parentheses are 95% confidence intervals.

Tab. 2. Numbers of complete twin pairs, concordant pairs, and discordant pairs, along with lifetime prevalence rates, probandwise concordance rates, and tetrachoric correlations for childhood speech-language disorders, stuttering, and cluttering, separately for monozygotic (MZ) and same-gender dizygotic (SSDZ) twin pairs, males and females

Trait	Zygosity*	Number of pairs	Number of concordant pairs	Number of discordant pairs	Prevalence	Probandwise concordance**	Tetrachoric correlation**
Males							
Speech-lang. disorders	MZ	649	42	79	0.13	0.52 (0.42,0.61)	0.79 (0.66,0.88)
Speech-lang. disorders	SSDZ	825	22	125	0.10	0.26 (0.18,0.36)	0.40 (0.18,0.58)
Stuttering	MZ	689	36	48	0.087	0.60 (0.49,0.70)	0.85 (0.71,0.93)
Stuttering	SSDZ	880	6	123	0.077	0.09 (0.03,0.18)	0.071 (-0.24,0.37)
Cluttering	MZ	684	48	118	0.16	0.45 (0.36,0.53)	0.62 (0.45,0.75)
Cluttering	SSDZ	875	20	160	0.11	0.20 (0.13,0.29)	0.31 (0.08,0.51)
Females							
Speech-lang. disorders	MZ	945	36	49	0.064	0.60 (0.48,0.70)	0.87 (0.76,0.94)
Speech-lang. disorders	SSDZ	1170	15	114	0.062	0.21 (0.12,0.31)	0.35 (0.12,0.55)
Stuttering	MZ	995	21	36	0.039	0.54 (0.39,0.67)	0.84 (0.67,0.93)
Stuttering	SSDZ	1207	4	92	0.041	0.08 (0.02,0.19)	0.15 (-0.19,0.46)
Cluttering	MZ	992	65	135	0.13	0.49 (0.41,0.57)	0.70 (0.57,0.80)
Cluttering	SSDZ	1209	33	216	0.12	0.23 (0.17,0.31)	0.32 (0.14,0.49)

*MZ = Monozygotic twins, SSDZ = Dizygotic twins from same-gender pairs.
**In parentheses are 95% confidence intervals.

MULTIVARIATE BIOMETRIC ANALYSIS

Type and magnitude of genetic and environmental influences on the traits were revealed by the multivariate liability-threshold model. Table 3 shows goodness-of-fit statistics of the full ADE model and hierarchical chi^2 tests of sub-models.

Tab. 3. Goodness-of-fit statistics of the full ADE model and hierarchical chi² tests of sub-models

Model	χ^2	df	p	c.t.m.	$\Delta\chi^2$	Δdf	p
1. Full ADE	80.640	57	0.021	---	---	---	---
2. AE	**100.637**	**69**	**0.008**	**1**	**19.997**	**12**	**0.067**
3. AE + no genetic correlations	587.569	75	0.000	2	486.932	6	0.000
4. AE + no unique environmental correlations	170.576	75	0.000	2	69.939	6	0.000

A = additive genetic factor; D = non-additive genetic factor; E = unique environmental factor.
χ^2 = Chi-square; df = degrees of freedom; p = p-value; c.t.m. = compared to model.
$\Delta\chi^2$ = difference in chi-square between nested models; Δdf = difference in degrees of freedom between nested models.

Table 4 shows the heritabilities and genetic correlations of childhood speech-language disorders, stuttering, and cluttering, as estimated under the best-fitting AE Cholesky decomposition. Substantial heritabilities for the traits were found. The estimates for males/females were 0.71/0.87 for childhood speech-language disorders, 0.78/0.80 for stuttering, and 0.53/0.65 for cluttering. The pattern of these estimates was very similar to that derived from the univariate analysis based on raw dichotomous data, which gave heritabilities of 0.79/0.86 for childhood speech-language disorders, 0.84/0.81 for stuttering, and 0.62/0.69 for cluttering in males/females (not shown). In the univariate analysis, observations from opposite-gender twin pairs also suggested that the same genes may be responsible for the effects in males and females in each disorder (not shown), though lifetime prevalences are different between genders. As a consequence, multivariate analysis was performed by fixing, for twins in OSDZ pairs, additive genetic correlation to 0.5 and non-additive genetic correlation to 0.25.

High genetic correlations between the traits emerged. The estimates for childhood speech-language disorders and stuttering were 0.71/0.79 for males/females, for childhood speech-language disorders and cluttering were 0.73/0.56, and for stuttering and cluttering were 0.53/0.57. Substantial unique environmental correlations between the traits were also obtained in both genders.

Tab. 4. Genetic and environmental proportions of variance and correlations as estimated, for males and females, under the best-fitting (AE) Cholesky decomposition for childhood speech disorders, stuttering, and cluttering

	Additive genetic (A) and unique environmental (E) proportions of variance*		
	Males		
	Speech-language disorders	Stuttering	Cluttering
A	0.71 (0.61,0.81)	0.78 (0.70,0.86)	0.53 (0.42,0.64)
E	0.29 (0.19,0.39)	0.22 (0.14,0.30)	0.47 (0.36,0.58)
	Females		
	Speech-language disorders	Stuttering	Cluttering
A	0.87 (0.81,0.93)	0.80 (0.70,0.90)	0.65 (0.57,0.74)
E	0.13 (0.07,0.19)	0.20 (0.10,0.30)	0.35 (0.26, 0.43)
	Additive genetic (upper triangle) and unique environmental (lower triangle) correlations*		
	Males		
	Speech-language disorders	Stuttering	Cluttering
Speech-language disorders	---	0.71 (0.63,0.78)	0.73 (0.60,0.86)
Stuttering	0.92 (0.76,0.99)	---	0.53 (0.40,0.68)
Cluttering	0.29 (0.07,0.50)	0.63 (0.38,0.85)	---
	Females		
	Speech-language disorders	Stuttering	Cluttering
Speech-language disorders	---	0.79 (0.71,0.86)	0.56 (0.47,0.66)
Stuttering	0.86 (0.52,0.99)	---	0.57 (0.45,0.68)
Cluttering	0.79 (0.54,0.95)	0.35 (0.05,0.64)	---

*In parentheses are 95% confidence intervals.

DISCUSSION

The most important finding in this study is genetic pleiotropy of childhood speech-language disorders, stuttering, and cluttering, as revealed by the high genetic correlations between the disorders, especially childhood speech-language disorders and stuttering, and childhood speech-language disorders and cluttering. This suggests that additive genetic effects may be largely shared by the disorders, and thus may play an important role in their co-morbidity.Both probandwise concordance rate and tetrachoric correlation were higher in MZ than in DZ pairs irrespective of gender, clearly pointing to genetic influences on behavior liability.

For stuttering, our 95% confidence interval for univariate heritability estimates are within the limits also found by Andrews et al. (1991), Felsenfeld et al. (2000) and Ooki (2005), who estimated the genetic proportion of variance in behavior liability at about 70%. A weaker, yet substantial, genetic contribution was found for cluttering, with heritability estimates of 62% in males and 69% in females. This is contrary to what Daly (1986) maintains from other authors, who suggested that the genetic component may be larger in cluttering than in stuttering. To explain this, we assume that our self reported "cluttering" are more widespread than traditional diagnosed cluttering. Cluttering is generally not very often diagnosed, and we may also assume that our self-reporting group include a broader definition of cluttering, such as the Cluttering Spectrum Behavior (CSB), proposed by Ward (2006) and used by the general public for those speakers who display some cluttering characteristics.

For extreme deficits in measures of productive vocabulary in a large epidemiological sample of selected twin pairs, Dale et al. (1998) reported a group heritability of 73%. Bishop et al. (1995) showed group heritability close to 100 % for deficits in measures of expressive and receptive language ability in their twin sample. Bishop & Haylou-Thomas (2008) demonstrated, in a new study, that the heritability may vary depending on diagnostic criteria; more precisely, low heritability is likely to be found for language disorders identified by population screening, while high heritability may emerge in children with speech-language difficulties referred for intervention.

It is noteworthy that, for each disorder, although lifetime prevalence rates differed between males and females, the same genes were suggested to affect liability in both genders. This draws attention to gender differences in gene-environment interaction, not modelled here.

The co-existence of cluttering with stuttering has been reported since Weiss (1935). Later, Weiss (1967) also suggested that stuttering might be grafted on cluttering secondarily, because stuttering generally started with cluttering-like symptoms and left a cluttering-like residue when cured.

Freund (1952) reported an increase of the incidence of abnormal speech hastiness among stutterers prior to and around puberty from 15 to 25 percent, and Preus (1981) reported 32 percent of people who stuttered also showed symptoms of cluttering; a finding Daly (1993, 1994) increased to approximately 40 percent. Seeman (1974) hypothesized a common genetic factor for stuttering and cluttering. Our estimates of a substantial genetic correlation between the two disorders are consistent with this hypothesis, but suggest that the shared heritable component is likely to be supplemented by genetic factors that are specific to stuttering or cluttering.

Finally, our results indicate that the heritability may be greater for stuttering than for cluttering, and that the two disorders may share less genetic factors with each other than with childhood speech-language disorders.

In addition to genetic effects, unique environmental effects contribute a moderate but significant influence on the expression of stuttering. Because common environmental influences, such as excessive parental concern about imperfect speech, a competitive and perfectionistic parental style, and a family drive for upward mobility have been implicated in stuttering etiology for several decades (cf. Johnson, 1959; Guitar, 2006, p.116-7), the non-significant common environmental parameter is of particular interest. So, the present study does not give any support for Johnson's "diagnosogenic theory" on stuttering etiology.

Environmental influences in childhood speech-language disorders have not been widely studied in twin samples. Van Hulle et al. (2004) stressed the importance of the shared environment, responsible for 53–77% of the variation in both boys and girls, in toddler expressive language development. They noted that unshared environment includes both environmental factors that are unique to each individual and measurement error, and they found that unshared environment accounted for 20% of the variance in boys and 13% of the variance in girls for two-word combination use, and less for other language modalities in toddlers at age 20-38 months. A recent study of Hayiou-Thomas (2008) shows unshared and shared environmental proportions of variance in the speech factor of 0.29 and 0.15, respectively. The value 0.29 for the contribution of unique environment was also found in our study for males.

To our knowledge, the environmental influences on cluttering development have not been studied in details, and very little knowledge is also available about the gene-environment interaction in childhood speech-language disorders, stuttering and cluttering.

Furthermore, our self-reported lifetime prevalences for childhood speech-language disorders are within the range of other published data, because indications from 5 to 8 per cent for children in the pre-school age are common in general, not specifically for twins. According to the National Institute on Deafness and Other Communication Disorders (NIDCD, 2011), the prevalence of speech sound disorder in young children is 8 to 9 percent. By first grade, roughly 5 percent of children have noticeable speech disorders. As regards gender difference, prevalence is generally twice to three times higher for boys compared with girls. Thus, our results showing higher prevalence rates in males are in accordance with the rates observed in singletons. The self-reported lifetime prevalences of stuttering, from less than 4 percent for females to 9 percent for males, are only slightly higher than those previously reported.

There are very few solid research data on cluttering (St. Louis, Raphael, Myers & Bakker, 2003). According to Brady (1993), cluttering is much rarer than stuttering. The highest prevalence of cluttering in the literature is given by Becker & Grundmann (1970) who found 9 clutterers among 606 pupils (1.5%) at the age of 7–8 years. These indications are in contrast with the values of 11 to 16 percent obtained in our self-report study. The almost equal ratio between males and females found here is also different from the male:female ratio of 4:1 shown by Arnold (1960). Yet, based on preliminary research (Raphael et al., 2005, St. Louis & McCaffy, 2005), St. Louis, et al. (2007) are uncertain that the 4:1 ratio will be confirmed by future research. We assume that the high prevalence of cluttering in our twins is related to a broader definition, Cluttering Spectrum Behavior (CSB), proposed by Ward (2006) and used by the general public for those speakers who display some cluttering characteristics. St. Louis, et al. (2007) also raise the possibility that cluttering may be underdiagnosed because relatively few clinicians are as knowledgeable about cluttering as they are about stuttering, few clutterers self-refer for services, and some clutterers do not believe they have a speech problem, and hence they do not seem to have a concern. St. Louis et al. (2010) indicate self-identification of cluttering to 8.9 percent among 90 Turkish respondents with a mean age of 31 years and a male/female ratio of 48–52%.

Our probandwise concordance rates for childhood speech-language disorders (52-60% for MZ, 21–26% for DZ) are lower than those found in the age range of five to sixteen years by Lewis & Thomson (1992), Bishop et al. (1995), Tomblin & Buckwalter (1998) and DeThorne et al. (2006). Here the ranges were 0.86 to 0.96 for MZ and 0.44 to 0.69 for DZ. Recently, Bishop & Hayiou-Thomas (2008) analyzed data from the Twin Early Development Study (Hayiou-Thomas, Oliver & Plomin., 2005), and they concluded that genetic studies are more likely to find high concordance rates and heritability if they focus on cases who have speech difficulties and who have been referred for intervention.

VALIDITY, STRENGTHS AND LIMITATIONS

The use of large population-based data from twins is a clear strength of the present study compared to investigations based on clinical samples. However there can be a number of drawbacks associated with using self-reported childhood speech-language disorders as a categorical definition in genetic studies. For every single twin the discrepancy level is arbitrarily defined relative to distributions in normal populations and without any

clear diagnostic criterion. Furthermore there are many subgroups of childhood speech-language disorders and different factors may be a result of this self-evaluation and the memory of it. We will just mention that Bishop et al. (1999) found that expressive language disorders and articulation disorders in combination with expressive language disorders showed strong evidence of heritability but there is no significant genetic influence on the auditory-processing measure. In case different factors lead to language problems in twins and singletons, this undermines a basic assumption of the twin method. But both twins and singletons with childhood speech-language disorders do have a high rate of affected relatives, consistent with a genetic etiology. Twinning might slow down language development in early childhood, but does not significantly affect childhood language at school-age and beyond (Bishop et al., 1999). Self-reported data might also be prone to recall bias, especially when old twins are asked about their communication problems in childhood, and the recollections of the twins might have been influenced by what they remembered of their behavior, relative to that of their co-twins. We also know that human memory is fallible (Schacter, 1999) and thus the reliability of self-reported data is tenuous, because of:

- *Transience:* Decreasing accessibility of information over time.
- *Absent-mindedness:* Inattentive or shallow processing that contributes to weak memories.
- *Blocking:* The temporary inaccessibility of information that is stored in memory.
- *Misattribution*: Attributing a recollection or idea to the wrong source.
- *Suggestibility:* Memories that are implanted as a result of leading questions or expectations.
- *Bias:* Retrospective distortions and unconscious influences that are related to current knowledge and beliefs.
- *Persistence:* Pathological remembrances-information or events that we cannot forget, even though we wish we could.

Those are common problems of self-evaluation in all surveys. We also know that not all people will understand our questions in the same way, but we have tried to construct our questions so that most people will understand the questions in the way we intended. For wording the questions we consulted the Danish IT Centre for Education and Research, an agency under the Danish Ministry of Education. This agency has a special field of competence on implementation of questionnaire surveys, including wording of questionnaires, and due to the large number of responders we assume we have minimized the problems related to the questions.

In this study the prevalence was calculated from self-reported retrospective data from the responders, not by clinical or everyday observations. An intrinsic limitation of the twin design is that twins are slower in language development than singletons (Rutter & Redshaw, 1991), although this delay diminishes during childhood for twins of higher occupational status (Thorpe, 2006). Finally, in biometric modeling, we made a number of simplifying assumptions that were not tested. One such assumption is that there are only additive effects of genes and environment on the phenotypic variance. However, in practice, there may be interactive effects. These interactive effects become incorporated into the estimates of genetic liability (for interactions between common environmental and genetic risk) or unique environmental estimates (for interactions between unique environmental and genetic risk). Consequently, the estimates of genetic and environmental variance may include these types of genes by environment interaction variance.

Despite the limitations mentioned above, our results derived from a large cohort of twins may aid to the understanding of the biological and environmental contributions to the development of childhood speech-language disorders, stuttering, and cluttering, as well as their co-occurrence. Our study might therefore contribute to the design of effective interventions and treatments (Button et al., 2007). The findings give also support for early intervention in children from families having developmental communication disorders in order to minimize long term effects of developmental communication disorders. Also children with perinatal or postnatal individual environmental episodes might also be at risk for developmental communication disorders.

CONCLUSION

With the limitations related to adult self-reports, this study demonstrates substantial genetic influence on the traits of childhood speech-language disorders, stuttering, and cluttering, and mutual genetic relationship between them. Substantial unique environmental correlations between the traits were also found in both genders.

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STRESZCZENIE

Dziedziczność i genetyczne zależności jąkania, giełkotu i zaburzeń rozwoju języka w opisie osób dorosłych

Celem pracy było zbadanie wzajemnego związku między jąkaniem, giełkotem i rozwojowych zaburzeniami mowy i języka na poziomie genetycznym u dorosłych poprzez zadawanie pytań jednozadaniowych (*single-item*). Zamiarem było potwierdzenie, czy te pytania zgodne są z wynikami przesiewowych bądź diagnozowanych danych wejściowych. Jeśliby stwierdzono zgodność, autorzy chcieliby rozszerzyć model biometryczny o nowy aspekt związku między tymi trzema cechami rozwojowych zaburzeń komunikacji ustnej.

Wzajemny związek miedzy jąkaniem, giełkotem i rozwojowymi zaburzeniami mowy i języka

Literatura opisuje równoczesne występowanie jąkania i rozwojowych zaburzeń mowy i języka, jąkania i giełkotu oraz giełkotu i rozwojowych zaburzeń mowy i języka. Ponieważ obserwowano, że jąkanie i giełkot występują w rodzinach, przypuszcza się, że w grę wchodzi dziedziczność. Również giełkot współwystępuje z opóźnionym rozwojem mowy, zaburzeniami płynności mowy, tempa, jąkaniem, ADHD, zaburzeniami artykulacji, języka, specyficznymi trudnościami w uczeniu się, CAPD i apraksją mowy, ale te dwa zaburzenia płynności mowy są odrębne. Co więcej, dostępna literatura na temat różnicowania jąkania i giełkotu sugeruje, że zasadnicza różnica między tymi populacjami klinicznymi koncentruje się na stopniu przygotowania mówiącego do powiedzenia zamierzonych słów. Jąkający się wiedzą, co chcą powiedzieć, ale coś przeszkadza im w próbach wypowiedzenia różnych słów, natomiast osoby z giełkotem niekoniecznie wiedzą, co chcą powiedzieć, ale i tak mówią. Na tym podłożu można by stwierdzić jakiś rodzaj wspólnej genetycznej bądź środowiskowej patogenezy tych trzech cech. Aby zbadać możliwe wzajemne powiązanie między opisywanymi przez samych pacjentów cechami jąkania, giełkotu i rozwojowymi zaburzeniami języka i mowy zastosowano podwójną metodę. Podejście autorów polega na równoczesnej ocenie z użyciem modelu wielowymiarowego dziedziczności każdej z tych cech i genetycznej korelacji między nimi.

Rozwojowe zaburzenia mowy i języka

Seeman (1937) pierwszy uznał, że czynniki genetyczne są odpowiedzialne za różne opóźnienia w rozwoju języka, zaś Ingram (1959) zaobserwował genetycznie uwarunkowaną dziedziczność konkretnych rozwojowych zaburzeń mowy w dzieciństwie. Oceny dziedziczności dla poszczególnych upośledzeń językowych były 46,418 nierówne: wskaźniki zgodności probantów (*probandwise concordance rates*) u bliźniąt jednojajowych wynosiły 0.36–0.96, a u dwujajowych 0.20–0.69. Wpływ czynników genetycznych był liczbowo większy u chłopców niż u dziewczynek. Zgodność probantowa u bliźniaków (zgodność probantów u bliźniąt) w przypadku niskiej zdolności językowej pokazuje wyraźną prawidłowość, przy czym znacznie większa jest zgodność dla bliźniaków jednojajowych niezależnie od płci w porównaniu z bliźniakami dwujajowymi tej samej płci, co wskazuje wpływ genetyczny na ryzyko, że współbliźniaki probantów są też dotknięte zaburzeniem. Przy mierze sumarycznej średnia ważona dla probantowej zgodności bliźniaków wynosiła 0.86 dla bliźniąt jednojajowych i 0.52 dla bliźniąt dwujajowych tej samej płci. Istnieje zatem ryzyko rzędu 86%, że drugi bliźniak także ma niskie zdolności językowe. U bliźniaków dwujajowych ryzyko spada do 52%. Ogólnie biorąc, wpływ płci był mały, ale znaczący, i istniało wskazanie, że rola genów jest silniejsza u chłopców niż u dziewczynek.

Jąkanie

Z literatury wynika, że jąkanie ma także podłoże genetyczne, przy czym jest względnie więcej jąkających się wśród bliźniaków niż jedynaków. Wykazano większą zgodność w kategorii parowania i probantów u bliźniaków jednojajowych niż dwujajowych Większy względny wskaźnik jąkających się wśród bliźniaków niż jedynaków przypisuje się czynnikom dziedzicznym. Większe wskaźniki zgodności odnoszą się do par jednojajowych niż dwujajowych, przy wariancji 71% przypisywanej addytywnej wariancji genów.

W innych badaniach par dorosłych bliźniaków, w których jeden lub obaj członkowie deklarowali jąkanie, przy zastosowaniu analizy dwuwymiarowej, stwierdzono w 70% wariancje podatności na jąkanie ze względu na addytywny wpływ genów, a 30% przypisano niewspólnym (*non-shared*) czynnikom środowiskowym

Brak jednego locusu podatności na jąkanie

Jąkanie to zaburzenie wieloczynnikowe i wielogeniczne, z wieloma mającymi wpływ loci w genomie w interakcji genów ze środowiskiem.

Giełkot

Wedle literatury, w giełkocie znaczną rolę odgrywa dziedziczność, a sam giełkot występuje cztery razy częściej u mężczyzn niż u kobiet w porównaniu z innymi zaburzeniami języka. Sugeruje się, że są dwa rodzaje wpływu dziedziczenia: specyficzne i niespecyficzne. Specyficzne to przekazywanie zespołu giełkotu w rodzinach, gdzie jest dużo jąkających się i z giełkotem. Niespecyficzne dziedziczenie objawia się przekazywaniem ogólnego upośledzenia (*disability*) językowego – opóźnionej mowy, dyslalii, dysleksji czy dysgrafii. Wedle autorów giełkot nie jest powszechnie uważany za zaburzenie.

MATERIAŁY I METODY

Opracowanie oparto na rozległym badaniu zbioru bliźniaków z 2002 roku, zorganizowanym przez Duński Rejestr Bliźniaków, zawierającym informacje o na temat ich zdrowia, chorób i przyczyn śmierci.

Osoby badane i kwestionariusz

Zbadano, w jakim stopniu współistnienie jąkania, giełkotu i rozwojowych zaburzeń mowy i języka wynika z genetycznej podatności wspólnej tym trzem cechom. Rozesłano kwestionariusz papierowy do przekrojowej grupy badanych – 46,418 bliźniakom urodzonym w latach 1931–1982. Bliźniaków klasyfikowano jako jednojajowych (MZ=*monozygotic*), dwujajowych tej samej płci (SSDZ=*same sex dizygotic*) i dwujajowych różnej płci (OSDZ=*opposite sex dizygotic*) w oparciu o cztery pytania o podobieństwo fizyczne używane do określenia wielorodności. Kwestionariusz zawierał 119 pytań dotyczących funkcjonowania, niesprawności, zdrowia, chorób, wykształcenia, zawodu, wagi, wzrostu, palenia papierosów, spożywania alkoholu, stosunków rodzinnych, dzieci, płodności, myśli i emocji.

Kwestionariusz ten wypełniło jedenaście grup badawczych. Niektóre pytania były ogólne i niezwiązane z jednym konkretnym projektem. Dwanaście pytań odnosiło się do niesprawności i upośledzeń związanych ze słuchem, mową, językiem i czytaniem, łącznie z doświadczeniem osobistym i samodeklarowanym: zapaleniem ucha środkowego, problemami ze słuchem, noszeniem aparatów słuchowych, chorobą Meniere'a, szumem w uszach, kryptofazją, rozwojowymi zaburzeniami mowy i języka, jąkaniem, giełkotem, nabytymi zaburzeniami mowy, afazją i dysleksją. Dwanaście pytań dotyczyło podawania niesprawności i upośledzeń słuchu, mowy, języka i czytania, w tym doświadczenia osobistego i samodeklarowanych: zapalenia ucha środkowego, problemów ze słuchem, noszenia aparatów słuchowych, choroby Meniere'a, szumów w uszach, kryptofazji, rozwojowych zaburzeń mowy i języka, jąkania, giełkotu, afazji i dysleksji.

Trzech pytań użyto do samoidentyfikacji przez bliźniaków dziecięcych zaburzeń języka i mowy, jąkania i giełkotu. :

„Czy miałeś problemy z mową i językiem w dzieciństwie? Czy jąkasz się bądź jąkałeś się?"

„Czy jest (było) dla ciebie problemem, że mówisz tak szybko, że zacinasz się na jakichś słowach i opuszczasz sylaby (giełkot)?"

Naszym zamiarem było opisać dziedziczność osobiście doświadczanych problemów z komunikacją. Samoocena ma wady, ale pozwala uzyskać wrażenia od samego badanego i pozwala ocenić zachowania trudne do wywołania w otoczeniu klinicznym.

U większości osób, które doświadczyły rozwojowych zaburzeń mowy i języka, jąkania i giełkotu , było to pierwotne zaburzenie rozwojowe.

Analiza statystyczna

Dla każdej cechy przeprowadzono obliczenia zbiorcze razem z częstotliwością występowania zaburzenia przez całe życie według płci i wielorodności. Dla każdej cechy obliczono przybliżone wskaźniki zgodności probantowej i korelacje tetrachoryczne oddzielnie dla par jednojajowych i dwujajowych tej samej płci oraz dla kobiet i mężczyzn.

WYNIKI

Spośród 280 878 (10 439 par) bliźniaków jednojajowych, dwujajowych tej samej płci i dwujajowych różnej płci 1580 podało rozwojowe zaburzenia języka i mowy, 1080 jąkanie i 2361 giełkot. 612 bliźniaków podało rozwojowe zaburzenia mowy i języka oraz jąkanie, 669 rozwojowe zaburzenia mowy i języka, i giełkot, a 483 bliźniaki podały jąkanie i giełkot zaś 319 bliźniaków wymieniło wszystkie trzy cechy.

Nie zaobserwowano znaczących różnic między bliźniakami z pełnych i niepełnych par w odniesieniu do wieku, płci, wielorodności i częstości występowania choroby przez całe życie w przypadku zaburzeń i korelacji fenotypowych co do tych cech.

W tabeli 1. podano sumaryczną liczbę bliźniaków, którzy zgłaszali rozwojowe zaburzenia mowy i języka, jąkanie i giełkot oraz częstość występowania w ciągu całego życia tychże niesprawności według wielorodności i płci. Rozwojowe zaburzenia języka i mowy wraz z jąkaniem występowały znacznie częściej u mężczyzn niż u kobiet, natomiast w giełkocie zaobserwowano tylko nieco większą częstość u mężczyzn. W jąkaniu częstość występowania była zgodna dla bliźniaków jednojajowych i dwujajowych. Zaobserwowano nieznacznie większe częstości występowania rozwojowych zaburzeń języka i mowy oraz giełkotu u bliźniaków jednojajowych w porównaniu z dwujajowymi

Tabela 2. pokazuje oddzielnie dla par jednojajowych i tej samej płci par dwujajowych oraz dla mężczyzn i kobiet liczbę pełnych par zgodnych i niezgodnych wraz częstością występowania, wskaźnikami zgodności probantów i korelacji tetrachorycznych dla rozwojowych zaburzeń mowy i języka, jąkania i giełkotu.

Dla wszystkich cech zarówno wskaźniki zgodności, jak i korelacje tetrachoryczne były znacząco wyższe u par jednojajowych niż u par dwujajowych tej samej płci, co wskazuje na istotny wpływ genetyczny na indywidualną podatność na każde z tych zaburzeń. W jąkaniu się korelacje tetrachoryczne sugerowały ewentualne nieaddytywne skutki genetyczne.

Typ i rozmiar wpływu genów i środowiska na badane cechy pokazuje wielowymiarowy model progu podatności ukazany w tabeli 3. (statystyka dobroci dopasowania).

Tabela 4. pokazuje dziedziczność i genetyczne korelacje dziecięcych zaburzeń języka i mowy, jąkania i giełkotu ocenianych według najlepiej pasującego rozkładu Cholesky'ego AE. Stwierdzono istotne dziedziczenie badanych cech.

DYSKUSJA

Najważniejszym ustaleniem opracowania jest stwierdzenie genetycznego pleotropizmu rozwojowych zaburzeń języka i mowy, jąkania i giełkotu, na co wskazywały wysokie korelacje genetyczne między tymi zaburzeniami, szczególnie rozwojowymi zaburzeniami i jąkaniem, i rozwojowym zaburzeniami i giełkotem. Sugeruje to, że zaburzenia podlegają w dużym stopniu wspólnym addytywnym wpływom genów, i tym samym odgrywają ważną rolę w ich współwystępowaniu.

Zarówno wskaźnik zgodności probantów, jak i korelacja tetrachoryczna były większe w parach jednojajowych niż dwujajowych niezależnie od płci, wyraźnie wskazując na genetyczny wpływ na podatność zachowania.

Warto zaznaczyć, że dla każdego zaburzenia, chociaż wskaźniki występowania były odmienne dla kobiet i mężczyzn, zasugerowano te same geny jako wpływające na podatność (na zaburzenia) w obu płciach. Zwraca to uwagę na różnicę między płciami w interakcji geny–środowisko, czego tu nie przedstawiano.

Współistnienie giełkotu z jąkaniem odnotowuje się w badaniach od 1937 roku. Ten sam badacz sugerował w roku 1967, że jąkanie może się wtórnie „przeszczepiać" na giełkot, ponieważ jąkanie zazwyczaj zaczyna się od objawów podobnych do giełkotu, a po wyleczeniu utrzymują się pozostałości giełkotopodobne.

I wreszcie, nasze wyniki wskazują, że dziedziczność może być większa w przypadku jąkania niż giełkotu, a te dwa zaburzenia mogą mieć mniej wspólnych dla siebie czynników genetycznych niż rozwojowe zaburzenia języka i mowy.

Oprócz wpływów genetycznych, swoiste wpływy środowiskowe mają umiarkowane, ale znaczące oddziaływanie na wyrażanie się jąkania. Ponieważ częste wpływy środowiska, takie jak nadmierna obawa rodziców co do niedoskonałej mowy, rodzicielski styl konkurencyjności i perfekcjonizmu oraz dążenie rodziny do posuwania się wyżej na drabinie społecznej były podawane (Johnson, 1959, Gitar, 2006) w etiologii jąkania od kilkudziesięciu lat, szczególnie byłby interesujący wspólny nieznamienny parametr środowiskowy. Niniejsze opracowanie nie potwierdza „teorii diagnozogenicznej" Johnsona co do etiologii jąkania.

Wedle wiedzy autorów, nie zbadano jeszcze dokładnie wpływu środowiskowego na rozwój giełkotu, niewiele także wiadomo o interakcji genetyczno-środowiskowej w rozwojowych zaburzeniach mowy i języka, jąkaniu i giełkocie.

Prawomocność, mocne strony i ograniczenia

Użycie danych opartych na dużej populacji bliźniaków jest wyraźnie mocną stroną niniejszej pracy w porównaniu z badaniami klinicznymi. Natomiast samodeklarowanie zaburzeń jest słabą stroną, tym bardziej że pamięć ludzka jest zawodna, co wpływa na rzetelność wyników z samooceny: w grę wchodzą takie czynniki, jak: przemijalność w czasie, roztargnienie, zablokowanie pamięci, błędne przypisywanie przyczyn, podatność na sugestie, tendencyjne przeinaczanie, uporczywość utrzymywania się informacji w pamięci.

Mimo ograniczeń, wyniki opracowania uzyskane z dużej populacji bliźniaków mogą pomóc w zrozumieniu wpływu biologii i środowiska na rozwój rozwojowych zaburzeń języka i mowy, jąkania i giełkotu oraz wpływu na ich współwystępowanie. Publikacja może zatem przyczynić się do opracowania skutecznych interwencji i terapii. Wyniki potwierdzają także konieczność wczesnej interwencji u dzieci z rodzin mających rozwojowe zaburzenia komunikacji, aby zminimalizować długofalowe skutki tych zaburzeń w sferze komunikacji. Dzieci z indywidualnymi środowiskowymi epizodami okołoporodowymi czy poporodowymi mogą być również zagrożone rozwojowymi zaburzeniami komunikacji.

WNIOSKI

Uznając ograniczenia samoopisu (*self-report*) dorosłych, niniejsza praca dowodzi znacznego wpływu genów na cechy dziecięcych zaburzeń mowy i języka, jąkania i giełkotu oraz na wzajemny genetyczny związek między nimi. Stwierdzono również znaczące swoiste korelacje środowiskowe między tymi cechami w obu płciach