Gene- Environment Interaction in Speech and Language Development- A Case Study of Monozygotic Twins

[1]Maya Sanghi, [2]Sidheshwar Pandey & [3]Hetal Shah

Abstract

Heritability of speech and language disorders is not a simple, straightforward process. There is good evidence that genetic factors play a role in the etiology of speech and language impairment; twin data may help us arrive at a clearer conception of the phenotype as well as quantifying the extent of the genetic contribution. By investigating identical and non-identical twins, researchers have been able to deduce that genetic variability plays a big role in determining whether or not a child will have developmental language problems. The current scientific view is that neither genetics nor environment is solely responsible for producing individual variation, and that virtually all traits show gene-environment interaction. In identical twins, DNA is same and they are brought up in the same surroundings hence case studies of the identical twins help us to study the effect of genetics, environment or both. Case studies also help in better understanding about the nature of the disorder. Aims of the study were 1)To improve our understanding of the nature of the speech and language disorder in identical twins and thereby facilitate further research in this area.2)To share the experience of working with a pair of identical twins with concomitant speech and language disorders. For assessment, 3D Language Acquisition test, Linguistic Profile Test, Stuttering Severity Index, Stuttering Chronicity Prediction Checklist and Pure Tone Audiometry were done. Possible causes of stuttering in the twins under study may be the interaction of genetic predisposition and precipitating factors such as home environment, competitive pressure etc. This paper also highlights the need for the clinicians to be alert about the speech language problems twins are prone to and the need to work in close co-operation with the pediatricians to prevent the developmental communication disorders in this population.

Key Words: Genetics, Environment, SSI, LPT, 3DLAT

Heritability of speech and language disorders is not a simple, straightforward process. In complex behavioral traits, such as stuttering, it can be difficult to identify the contribution of gene or genes and their interaction with environmental factor. One or more genes may be responsible for the underlying susceptibility of stuttering and they may have wider effects and cause susceptibility to other disorders as well. They may also interact with other genes to cause variety of effects (Yairi, Ambrose & Cox, 1996). Conditions that sometimes coexist with stuttering, such as phonologic disorders, learning deficits, and lower intellectual function, may be explained in this fashion. There is good evidence that genetic factors play a role in the etiology of speech and language impairment; twin data may help us arrive at a clearer conception of the phenotype as well as quantifying the extent of the genetic contribution. By investigating identical and non-identical twins, researchers have been able to deduce that genetic variability plays a big role in determining whether or not a child will have developmental language problems.

Fraternal twins (commonly known as "non-identical twins") usually occur when two fertilized eggs are implanted in the uterine wall at the same time. The two eggs form two zygotes, and these twins are therefore also known as dizygotic as well as "biovular" twins. Dizygotic twins, like any other siblings, have an extremely small chance of having the exact same chromosome profile.

Identical twins occur when a single egg is fertilized to form one zygote (monozygotic) which then divides into two separate embryos. Although their traits and physical appearances are not exactly the same due to environmental conditions both in and outside the womb, they do have identical DNA. The two embryos develop into fetuses sharing the same womb. When one egg is fertilized by one sperm cell, and then divides and separates, two identical cells will result. If the zygote splits very early (in the first 2 days after

[1]*Lecturer AST Department, T.N.M.C. MUMBAI,* [2&3]*Students, Shyam Bhavan No.3, 5th Floor, Gazdar Street, Chira Bazaar, Mumbai-400002, email- hetal.aslp@gmail.com.*

fertilization) they may develop separate placentas (chorion) and separate sacs (amnion). These are called dichorionic, diamniotic (or "di/di") twins, which occurs 20-30% of the time (which is present in our case study). Most of the times in identical twins the zygote will split after 2 days, resulting in a shared placenta, but two separate sacs. These are called monochorionic, diamniotic ("mono/di") twins. In about 1% of identical twins the splitting occurs late enough to result in both a shared placenta and a shared sac called; monochorionic, monoamniotic ("mono/mono") twins (Nieuwint, Van Zalen-Sprock, Hummel, Pals, Van Vugt, Van Der Harten, Heins, & Madan, 1999).

Concordance as used in genetics usually means the presence of the same trait in both members of a pair of twins. However, the strict definition is the probability that a pair of individuals will both have a certain characteristic; given that one of the pair has the characteristic. For example, twins are concordant when both have or both lack a given trait. A twin study examines the concordance rates of identical twins having the same trait, especially a disease. This can help determine whether the disease has a genetic cause. Thus, the concordance rate of a given characteristic helps establish whether or to what extent it is caused by genetic mutation (Lewontin, 1982).

Twin language is unique in several ways. One of the aspects of the 'twin situation' is that each twin receives relatively less individually directed parental speech (Conway, Lytton, & Pysh 1980; Lytton, Conway, & Suave 1977; Stafford 1987; Tomasello, Mannle, & Kruger 1986).

Gene Environment interaction- Some researchers think that interaction between genes and environment, rather genes and environment separately, may influence many traits. The current scientific view is that neither genetics nor environment is solely responsible for producing individual variation, and that virtually all traits show gene-environment interaction (Yairi, Ambrose & Cox, 1996).

Need for the Case Report: In literature, researchers have used twin studies to try to disentangle the environmental and genetic backgrounds of a cornucopia of trait. Modern twin studies also try to quantify the effect of a person's shared environment (family) and unique environment (the individual events that shape a life) on a trait. Twin researchers acknowledge that limitations do exist, but they further add that the limitations don't negate the usefulness of twin studies. In identical twins, DNA is same (i.e. all genes are the same) and they are brought up in the same surroundings hence case studies of identical twins help us to study the effect of genetics , environment or both. Case studies also help in better understanding about the nature of the disorder. Further, single case study designs help us answer questions about the treatment effects. In India, to the best of our knowledge, there are very few studies (published in literature) on problems in speech and language development in identical twins. Hence the clinicians intended to report this study.

Aim: 1) To improve our understanding of the nature of the speech and language disorder in identical twins and thereby facilitate further research in this area.

2) To share the experience of working with (an interesting case reported to our department) a pair of identical twins with concomitant speech and language disorders.

There is a pervasive assumption in the twin language development literature that twins are somewhat delayed in language development and more prone to language disabilities.

Day (1932) studied 80 pairs of twins and 140 singletons. On several gross measures of language complexity (such as sentence length, number of different grammatical categories in a sentence) twins were found to be as much as two years behind singletons by the age of five.

Lytton (1980) and Conway, Lytton & Pysh (1980) report a study comparing twins and singletons on language measures. Their study suggests that both biological and environmental variables contribute to language delays in twins, and the authors conclude that the environmental variables are more important.

Lewis & Thompson (1992) studied fifty-seven same-sex twin sets (32 monozygotic and 25 dizygotic) for concordance of speech and language disorders. Results showed monozygotic twins to have higher concordance than dizygotic twins.

A half-dozen studies of twins have found that concordance for stuttering (both twins either stutter or don't stutter, rather than one twin stuttering and the other not stuttering) is much more likely in identical twins than in fraternal twins. Yairi, Ambrose & Cox (1996) also endorses the same findings.

A review of the literature regarding recovery from stuttering indicates that clinicians might expect that two out of every three stutterers observed in a school –age population will recover spontaneously. Recognizing a need to identify those who will not recover without intervention, and in view of the lack of research on which to

base such identifications, a stuttering chronicity prediction checklist for use in such research was devised. This inventory consists of 27 questions which the speech clinician may answer with yes or no responses after consultation with the stutterer's parent and after observations of and interaction with stutterer. "Yes" response may be interpreted as predictors of stuttering chronicity, but no individual question weighting for predictive value should be attempted. However, the validation of the checklist is not yet done (Cooper, 1973).

Dworzynski, Remington, Rijsdijk, Howell, & Plomin (2007) conducted a study to assess contribution of genetic factors in the persistence and early recovery from stuttering. The study also tried to address the question whether there are different genetic and environmental influences for children who recover from stuttering and those who persist. To answer this question data from the Twins Early Development Study were employed. Parental reports regarding stuttering were collected at ages 2, 3, 4, and 7 years, and were used to classify speakers into recovered and persistent groups. Thus, they concluded stuttering appears to be a disorder that has high heritability and little shared environment effect in early childhood and for recovered and persistent groups of children, by age 7.

Concomitant Speech and Language Problems in Children Who Stutter

Research has found that other speech and language difficulties more common among children who stutter than those who do not.

Another perspective of some authorities is that a common deficit may be responsible for the articulation errors, language deficits and stuttering and this deficit may be passed on genetically. Delayed development of circumscribed areas of the brain responsible for speech and language related functions might result in language, articulation and fluency difficulties combination. Some differences on i) How the brain processes such functions and ii) the kind of stimulation the child was exposed to, may tip the balance towards any one of these disorders or in any combination (Silverman,1992). Clinicians with this view would work intensively on all areas involved, simultaneously. Recent literature in this area supports the concept for integrating work on fluency with other speech and language problems in a child with beginning stuttering.

Case Study

Twins (same sex- male) named K.S and J.S. aged 5yrs 8 months were brought to our Audiology and Speech Therapy department with a complaint of stuttering.

Case History of Both Twins: No significant pre-natal history

Peri-natal History: Pre term (30 weeks) normal delivery. CIAB, BW- 1.56 Kg, Length-41 cm, HC-30.5 cm (Twin I), BW- 1.33 Kg, Length-38 cm, HC-29.5 cm (Twin II)

Post natal History: Both twins were kept in NICU for 28 days. ? Viral G. E. at the age of 2 yrs. (Twin I)

Medical History

- Diamniotic Dichorionic twins and each had 2 episodes of Apnea.
- APGAR Score: 1 – 7/10, 5- 8/10. (Twin I) , 1 – 6/10, 5- 7/10.(Twin II)
- Preterm male neonates with AGA with sepsis.

On Examination

- Eyes- Right eye-Squint present, Left eye - Vision affected(Twin I),
- Eyes – Normal(Twin II)

Motor Milestones were typical.

Speech Milestones: There is a history of delayed speech milestones in the family (Father and Grandfather). The children's history is also suggestive of delayed speech milestones.Vocalizations-1yr of age, Babbling-1.6 yrs of age,

1st word- 2.2 yrs of age, 3 word sentences-4 yrs of age.

Fluency Problem: Mother reported that the Twin I had history of dysfluency for a short period at the age of 3 yrs and there was spontaneous recovery. However, dysfluencies increased in the child at the age of 5.8 yrs and parents reported with the child for therapy. Twin II became markedly dysfluent around 3.4 yrs of age and took therapy for 3 months. Due to mother's health problems, they didn't attend therapy for about a year. Again dysfluencies increased at the age of 5 .8 yrs. Hence this child registered for therapy once again along with Twin I.

Family History: Father of the children had a history of mild stuttering in childhood and he recovered without any treatment. As reported by the parents, children's father's first cousin sister (maternal uncle's daughter) is a PWS. They stay in a joint family.

Handedness: Right handedness was noticed at the age of 2.5 yrs in both the twins. Twin I still erases with left hand. Twin II is predominantly right handed. Father is left handed only for writing.

Education: Both the twins are attending Sr. Kg. in school which has English as the medium of instruction.

Mother Tongue: Hindi, Marwari

Assessment

Language Assessment was carried out using 3D Language Acquisition Test (Vaidyanathan, 1989) for both the twins. Results revealed: Comprehension: 30-32 months, Cognition: 30-32 months, Expression: 27-29 months suggestive of delay of about 34 months (2 yr 10months) in comprehension and 37months (3 years 1 month) in expression.

Hearing Assessment: Pure Tone Audiometry revealed Bilateral Hearing Sensitivity within Normal Limits for both the twins.

Muscle tone was assessed by physiotherapist and it was seen that their muscle tone is normal. (This was ensured in view of children who are preterm are susceptible for muscle tone abnormalities).

Intelligence Quotient Assessment: Results revealed Average Intelligence (on Seguin Form Board) in both the twins.

The Cooper Chronicity Prediction Checklist (Cooper,1973) for School Age Stutterers- Twin I-11/27 , Twin II-10/27 which predicts that children may have a good chance of outgrowing stuttering.

Stuttering Severity Index (Riley, 1980): The scores were calculated on spontaneous speech and picture description task. To check the reliability of scores obtained two clinicians separately calculated the scores of the same sample. To check the consistency of the disorder and its degree the test was administered twice first in session1 and then after two weeks in session 3.

TWIN I- The scores revealed Severe Stuttering.

Twin II- The scores revealed Moderate Stuttering.

Mother- child interaction was assessed and it was observered that the mother is very authoritative, perfectionist and uses complex sentences while conversing. To check the anxiety and depression levels in the parents Hospital Anxiety And Depression Scale (Snaith and Zigmond, 1994) was administered on both parents. It revealed that Mild Anxiety is present in the father.

Management

Long Term Goal: To achieve age appropriate language skills and to achieve fluent speech in all situations within the limit of children's potential.

Short Term Goals

1) Parent counseling.

2) Language Therapy: Variety of activities was implemented to enhance comprehension and expression.

3) Activities for Fluency Enhancement were carried out.

4) Since there was competition for speaking between the twins which in turn increased the dysfluencies hence turn taking was encouraged.

5) Most often, activities for fluency and language were incorporated in the games. Children enjoyed receiving reinforcement and hence were motivated to use slow rate of speech and simple complete sentence.

6) Parents were an active part of the therapy sessions and were therefore advised to follow similar activities at home.

7) Parents tried their best in spite of time constraints, as they were very motivated.

Assessments After 18 Sessions: On Linguistic Profile Test scores (for language assessment) revealed:

Twin I: Receptive Language Age- 4-4½ yrs, Expressive Language Age-3 ½-4 yrs that is language delay of about 1 ½ yrs in comprehension and 2 yrs in expression as compared to earlier delay of 2 yrs 10 months in comprehension and 3 yrs 1 month in expression.

Twin II: Receptive Language Age- 4 ½ -5 yrs, Expressive Language Age-4-4 ½ yrs that is language delay of about 1 yrs in comprehension and 1 ½ yrs in expression as compared to earlier delay of 2 yrs 10 months in comprehension and 3 yrs 1 month in expression.

SSI scores for stuttering revealed Twin I – Mild Stuttering (Total Task Score=12), earlier result revealed Severe Stuttering score being 25.

Twin II- Mild Stuttering (Total Task Score=10), earlier result revealed Moderate Stuttering score being 21.

Perceptually both the children are showing significant reduction in hard attacks and physical concomitants. Parents also feel that stuttering has decreased in both the children.

Discussion and Conclusions

Language Delay: There is a family history of delayed speech and language development (grandfather and father). In the twins in the present study, the Language delay was 2 yrs 10 months in

comprehension and 3 yrs 1 month in expression on the initial assessment.

This endorses the study done by Day (1932) that twins were found to be as much as two years behind singletons by the age of five.

Language delay in the present study could be due to:

i) Twining and genetic predisposition. ii) Inadequate language stimulation at home.

iii) Bilingualism: Since children are exposed to Hindi language at home but in their school, medium of instruction is English.

The possible explanations for stuttering present in the children are

Genetic Predisposition – There is family history of fluency disorder. Father of the children had a history of mild stuttering in childhood and from which he has recovered without any treatment. Father's first cousin sister (maternal uncle's daughter) is a stutterer as reported by the parents of the children.

Environmental Factors: The precipitating factors present in the environment are- i) Both parents are extremely busy, therefore there was lot of time pressure imposed on the children during speaking activities. It was also observed parents had fast rate of speech.

ii) Mother is very authoritative, perfectionist and uses complex sentences.

iii)Bilingualism.
iv)There are demands on the children in terms of display of speech. v) Competition for the floor to speak amongst the twins.

Thus our study supports the view that it is not the genetic or the environmental factor alone that plays a role in speech and language disorders but an interaction of both. Similar view was given by Felsenfeld, Kirk, Zhu, Statham, Neale, & Martin (2000).

Good chance of recovery? The following facts are suggestive of these children having very high probability of recovery from stuttering.

Firstly, these children reported at early age for therapy, particularly Twin II. Bentley (1988) studied the speech and language development of twins and one of the conclusions drawn by him is that the earlier the speech and language delays are identified, the better are the chances of successful treatment. Our experience also endorses this finding as children are responding well to the treatment program so far.

Secondly, the Chronicity Prediction Checklist scores -11/27(Twin I) and 10/27 (Twin II) that is less than 50% of the total scores, are suggestive of both the children having good chances of outgrowing stuttering (Lesser the scores better is the outcome).

The third fact is that the father of the children had outgrown his mild stuttering and therefore chances that these children may outgrow stuttering are high. This can be said on the basis of recent data indicating a strong genetic factor in recovery from stuttering. Both persistent and recovered stuttering tends to run in families (Yairi et al., 1996). A study done by

Dworzynski et. al (2007) revealed that a positive family history, and the fact that both early recovery and persistence are heritable, may play a role in risk assessment. Therefore genetic information can be employed at diagnostic, prognostic, treatment, and counseling levels.

Thus, there are high chances that children may outgrow fluency disorder. Clinicians are optimistic about their outgrowing the fluency disorder since children have responded well to the therapy so far. However to confirm our belief children should be kept on long term follow up.

This paper also intends to emphasize that many a times a clinician is likely to miss the concomitant speech and language disorders that occur along with stuttering. Parents themselves may not be aware of coexisting disorders and may come with the sole complaint of fluency problem. However one must not over look the importance of detailed assessment in all the domains of speech as well as language, however busy the clinician may be.

The studies reported by Day (1932), Lewis and Thompson (1992) , Yairi et al. (1996) suggest that it would be beneficial to keep all the twins on a regular follow up for assessment and management (if required) of their speech and language problem from an early age and thus, facilitate secondary prevention of speech and language challenges they are prone to. It would be important that pediatrician and speech language pathologist work hand in hand to prevent the speech and language problems in twins.

References

Bentley, P. (1988). Speech and Language Delays in Identical Twins. Retrieved September 8, 2007 from Web site: http://www.eric.ed.gov

Conway, D., Lytton, H., & Pysh, F. (1980). Twin-singleton language difference. *Journal of Behavioral Sciences, Vol.12*, 264-271.

Cooper, E. (1973).The Development of a Stuttering Chronicity Prediction Checklist: A Preliminary Report, *Journal of Speech and Hearing Disorders*, *Vol. 2,* 215-221.

Day, E. J. (1932). The development of language in twins 1. A comparison of twins and single children 2. The development of twins: their resemblances and differences. *Child Development, 3*: 179-199, 298-316.

Dworzynski. K., Remington .A., Rijsdijk. F., Howell. P. & Plomin. R. (2007). Genetic Etiology in Cases of Recovered and Persistent Stuttering in an Unselected, Longitudinal Sample of Young Twins. *American Journal of Speech-Language Pathology*, *Vol.16*:169-178.

Felsenfeld, S., Kirk, KM., Zhu, G., Statham, DJ, Neale, MC & Martin NG. (2000). A study of the genetic and environmental etiology of stuttering in a selected twin sample. *Behavioral Genetics, 30:*359–366.

Guitar, B. (1991). *Introduction to stuttering*; *Nature of stuttering*, Stuttering: An integrated approach to its nature and treatment; Baltimore, London, 2 ed; Lippicot Williams and Wilkins.

Lewis, & Thompson, J. (1992). A study of developmental speech & language disorders in twins. *Speech and hearing res., Vol. 35*: 1086-1094.

Lewontin, R. (1982). Human Diversity. Retrieved September 19, 2007 from Web site: http://en.wikipedia.org/wiki/Concordance_%28genetics%29

Lytton, H. (1980). *The socialization process observed in twin and singleton families*. Parent- child interaction, Plenum, New York.

Lytton, H., Conway, D., & Sauve, R. (1977). The impact of twinship on parent-child interaction. *Journal of Personality and Social Psychology, Vol. 25*: 97-107.

Nieuwint, A., Van Zalen-Sprock , R., Hummel, P., Pals, G., Van Vugt, J., Van Der Harten, H., Heins, Y., & Madan, K. (1999). "'Identical' twins with discordant karyotypes". *Prenatal Diagnosis*. *Vol. 19 (1):* 72-76

Riley(1980), Stuttering Severity Index.

Snaith, R. D. & Zigmond, A. S. (1994). Hospital Anxiety And Depression Scale. *Acta Psychiatra, Scand*, *Vol. 67*: 361 – 370.

Stafford, L. (1987). Maternal input to twin and singleton children: Implications for language acquisition. *Human Communication Research, Vol. 13*: 429-462.

Silverman, F. (1992). *Etiology of Stuttering*, Stuttering And Other Fluency Disorders, New Jersey, Prentice and Hall, pp.117-123, 226.

Souter, V., Parisi, M., Nyholt, D., Kapur, R., Henders, Opheim, K., Gunther, D., Mitchell, M., Glass, I., & Montgomery, G. (2007). A case of true hermaphroditism reveals an unusual mechanism of twinning, *Human Genetics journal, Vol. 121: No 2:* 179-185.

Tomasello, M., Mannle, S. & Kruger, A. (1986). Linguistic environment of 1- to 2-year-old twins. *Developmental Psychology*, *Vol. 22*: 169-176.

Vaidyanathan, R. (1989). Language Developmental Scale for Pre-school Children: A pragmatic approach. *Interdisciplinary seminar on language process and language disorder.* Osmania University. Hyderabad.

Van Riper, C. (1982). *Organicity of Stuttering,* The Nature Of Stuttering, Toronoto, Prentice and Hall 2 ed.

Yairi, E., Ambrose, N., & Cox. N. (1996) Genetics of Stuttering; A Critical Review. *Journal of Speech and Hearing Research, Vol39:* 771-784.