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# EARLY COMMUNICATION DEVELOPMENT IN RETT SYNDROME

BY

SHELLEY TAMS-LITTLE

A thesis submitted to the Faculty of Graduate Studies and Research in partial fulfilment of the requirements for the degree of Master of Science.

in

SPEECH - LANGUAGE PATHOLOGY

DEPARTMENT OF SPEECH PATHOLOGY
AND AUDIOLOGY

Edmonton, Alberta
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FACULTY OF GRADUATE STUDIES AND RESEARCH

THE UNDERSIGNED CERTIFY THAT THEY HAVE READ, AND RECOMMEND TO THE FACULTY OF GRADUATE STUDIES AND RESEARCH FOR ACCEPTANCE, A THESIS ENTITILED **EARLY COMMUNICATION DEVELOPMENT IN RETT SYNDROME**
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Supervisor, Dr. G. Holdgrafer, Ph.D.

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**ABSTRACT**

A survey study was employed to collect information on early communication development and regression in children with Rett Syndrome. Seventeen retrospective surveys were completed by parents of children with Rett syndrome between the ages of 23 to 48 months. Twelve of the 17 parents completed the questionnaire a second time at least three weeks later to determine parent reliability across two completions of the retrospective survey.

A few questions on the questionnaire requested parents to recall general information about gross and fine motor development, eye-contact, play, and atypical hand movements. The majority of the questionnaire involved early prelinguistic and linguistic communication development employing the Clinical Linguistic And Auditory Milestone Scale and other supplemental material. Results indicated that most of the 17 children with Rett syndrome exhibited an abnormality in communication development from birth or infancy. A few children were delayed from the onset with an abrupt early arrest in communication development. The most predominant pattern however was scattered and delayed development toward a highest attained milestone level of approximately 6 to 16 months prior to regressing. In addition most children did not evidence the communicative gestures which generally develop between age 9 to 13 months. The results suggest that the debilitating effects of the syndrome have an

impact at an early age.

Dedicated to and with many thanks for the support from my parents Marianna & Mel, Cy & Donna, and my grandparents.

A thank you to my University of Alberta professors and to my employers at the Red Deer Regional Health Unit.

A warm thanks to the volunteers at the Canadian and International Rett Syndrome Associations.

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Abbreviations:

RS=Rett syndrome

mos.=months

S.D.= standard deviation

at.h.m.= atypical hand movements

# CHAPTER 1

## Introduction

### Early Communication Development In Rett Syndrome

Rett syndrome (RS) is often referred to as a progressive neurological disorder (Badr, Witt-Engerstrom, & Hagberg, 1987; Budden, 1986; Holm, 1985; Pelson & Budden, 1987; Rosemberg, Arita, & Campos, 1986; Verma, Chheda, Nigro, & Hart, 1986), or a progressive encephalopathy (Walhstrom & Anvret, 1986). Presently, it is found exclusively in females (Coleman, Pines, & Bias, 1987; DeGennaro, McCaffery, Kirchgessner, Yang-Feng, & Francke, 1987; Donnelly, 1986; Goutieres & Aicardi, 1986; Hagberg, 1985a; Hagberg, Aicardi, Dias & Ramos, 1983; Hagberg, Goutieres, Hanefeld, Rett, & Wilson, 1985; Hillig, 1985; Kulz, Pohl, & Schober, 1985; Murphy, Naidu, & Moser, 1986; Trevathan & Naidu, 1988; Walstrom, 1987).

Rett syndrome became known at the international level in the 1980's (Haas, 1988; Lugaresi, Cirignotta, & Montagna, 1985), and in 1984 a diagnostic criterion was established at the Rett syndrome conference in Vienna (Hagberg et al., 1985; Trevathan & Naidu, 1988).

A number of investigations have been published focusing on the etiology of RS; however, the cause remains unknown (Al-Mateen, Philippart, & Shields, 1986; Bachmann, Colombo, Gugler, Kilian, Rett, & Silva, 1986; Badr et al.,

1987; DeGennaro et al., 1987; Donnelly, 1986; Hillig, 1985; Leiber, 1985; Nomura, Honda, & Segawa, 1987; Rett, 1986; Rolando, 1985; Tariverdian, Kantner, & Vogel, 1987; Rosemberg et al., 1986).

In reviewing the existing literature it appears that information is lacking concerning RS children's communication development and regression. Existing case studies have commented on communication development as normal initially and then regressing to a severely handicapping condition by age two or three years. The gap in information is particularly apparent for the first two to three years of a RS child's life. Hagberg and Witt-Engerstrom (1986) stated that the classical Rett syndrome picture is easily recognized at ages four to five; however, the period during infancy, with its initially vague signs, is still easily misinterpreted. Study of early communication development in children with RS may provide important additional information.

Details of prelinguistic and early linguistic development will become increasingly important if an etiology or medical treatment becomes known. Information on communication development may provide the early groundwork for further studies focusing on facilitating earliest possible detection and providing guidelines to monitor the child's development. Hagberg and Witt-Engerstrom (1986) suggested if an etiology and medical

treatment becomes known there would be a need for a screening tool combining the assessment of communication abilities and fine manipulative hand skills. Though early medical detection prior to observed symptoms/delays would be a better scenario. Once the regression reaches the point of severe mental retardation, reversing the regression would be unlikely (Rett, 1986).

Whether a treatment preventing the regression is discovered or not, further information for communication milestones/regression may in the long run facilitate the professional's ability to counsel parents. Parents could then be better informed about expectations for their child's regression and development. Budden (1986) stated a clearer understanding is necessary to relieve parents from feelings of guilt and inadequacy, and to provide for realistic goals in the management of their children.

The relevance of further study of this syndrome is crucial to health services in light of the prevalence of RS. The prevalence is estimated as twice as common as phenylketonuria (PKU) (Hagberg 1985a, 1985b). Prevalence studies have been completed in Sweden (Hagberg, 1985a, 1985b), Scotland (Kerr & Stephenson, 1986), and Italy (Zappella & Cerioli, 1987). The prevalence mentioned in many articles is Hagberg and Witt-Engerstrom's (1987) estimate of 1.0/10,000 live births. Hagberg (1985a) hypothesized the syndrome may be responsible for 1/4 to

1/3 of the cases of severely mentally handicapped girls.

The objective of this proposed investigation is to collect information on prelinguistic and early linguistic communication development to provide additional information to describe characteristics of the syndrome. This information may contribute to the theoretical model of stages of the syndrome's progression, and may have an impact on future clinical work and research.

# CHAPTER 2

## Review of the Literature

Rett syndrome was first described in a journal published in German in 1966 (Haas, 1988; Lugaresi et al., 1985; Moser, 1985), but did not become known at the international level until the early 1980's (Haas, 1988; Moser, 1986). Two explanations were offered concerning the 17-year delay in world wide recognition of RS. The early 1966 article failed to attract recognition, because it was published in German rather than English and because the journal the article was published in was not well read at the international level (Moser, 1986). Hagberg et al.'s (1983) report of 35 patients from Sweden, France, and Portugal in an American neurological journal introduced RS to the English-language medical literature (Adkins, 1986; Haas, 1988; Trevathan & Adams, 1988). This 1983 article spurred an awareness of RS at the international level (Haas, 1988; Rett, 1986). The 1980's have been a decade of growth with many published research articles and regularly held international Rett syndrome conferences (Haas, 1988).

A number of investigations have been published focusing on the etiology of RS. Research has been directed to the following areas: metabolic, biochemical,

anatomical, physiological, neuropathological, clinical manifestations, and genetics. Though there has been a proliferation of studies investigating the cause of Rett syndrome, the etiology remains unknown (Al-Mateen et al., 1986; Bachmann et al., 1986; Badr et al., 1987; D[illegible]ennaro et al., 1987; Donnelly, 1986; Hillig, 1985; Leiber, 1985; Nomura et al., 1987; Rett, 1986; Rolando 1985; Rosemberg, 1986; Tariverdian et al., 1987 ). Several case studies reported that children with RS were often misdiagnosed (Adkins, 1986; Al-Mateen, et al., 1986; Budden, 1986; Gillberg, 1987; Goutieres & Aicardi, 1987; Hagberg & Witt-Engerstrom, 1986; Lieb-Lundell, 1988; Missliwetz & Depastas, 1985; Olsson, 1987; Olsson & Rett, 1985; Percy, Zoghbi, Lewis, & Jankovic, 1988; Zapella, 1985). Studies frequently reported an incorrect diagnosis of autism, but the following conditions or labels were also reported: cerebral palsy, primary central nervous system malformation, mental handicap, primary seizure disorder, encephalopathy of uncertain etiology, and acquired encephalopathy.

A diagnostic criterion (Appendix A) was established by the consensus of physicians at the Rett syndrome conference in Vienna in 1984 (Hagberg et al., 1985; Trevathan & Naidu, 1988). The diagnosis relies on clinical observation and is tentative until ages two to five. A child must meet all of the eight criteria before

she can be diagnosed as typical or classical RS. Children meeting some of the diagnostic criteria, but not all eight, cannot be given the diagnosis of classical or typical Rett syndrome. Instead the incomplete Rett syndrome cases are termed "forme fruste", "incomplete", "atypical" or "variant" cases (Goutieres & Aicardi, 1986; Hagberg & Rasmussen, 1986; Hagberg & Witt-Engerstrom, 1986; Percy, Zoghbi, & Glaze, 1987; Zapella, 1985). Once an etiology is known it is possible the forme fruste cases may actually be confirmed as Rett syndrome (Goutieres & Aicardi, 1986).

Further information concerning Rett syndrome is desperately needed, but researchers have cautioned that information should not be added to the present criteria if it narrows diagnosis (Burd, & Gascon, 1988; Opitz, 1986). Opitz (1986) reported that because the cause is not known, it is impossible to ascertain the true degree and the complete extent of variability. If the criterion becomes too strict there is the risk of under-diagnosis.

Kerr, Montague, and Stephenson (1987) stated the significance of timing and style of onset of the clinical signs in Rett syndrome are of great significance. There is variability in the age of onset of certain symptoms; therefore, it is the sequence and relationship of development and symptoms that is central to the understanding of Rett syndrome (Kerr et al., 1987).

The development of communication abilities can not be fully understood unless other areas of development are reviewed as well.

Hand-Movements

Observable hand movements are recognized as an important variable to include in the study of RS. A description of hand movements was included in Hagberg and Witt-Engerstrom's (1986) first two stages: stage I, handwaving-unspecific and episodic; stage II, loss of hand use or skill, loss of acquired purposeful use of the hands, and handwringing, clapping, and washing stereotypes between ages one to four.

It was the hand movements that were first recognized by Dr. Rett (Moser, 1986). Moser (1986) reported Dr. Rett observed two girls in his waiting room, both displaying exactly the same hand washing behaviour that he had observed previously in six other clients. Dr. Rett felt the stereotypical movements were distinctive and differed from other handicapped children's stereotypical movements (Moser, 1986). Olsson (1987) stated that the most characteristic stereotypes in RS children are the hand washing movements with the hands together, arms flexed, in front of the chest or chin. Witt-Engerstrom (1987) reported that the loss of acquired manipulative hand skill, often mixed with stereotypical circulating hand-mouth movements, still constitutes the single most

informative warning signal of RS. Budden (1986) reported that in the absence of biochemical or chromosomal markers, the hand movement stereotypes can be used to distinguish this syndrome from other mental retardation, cerebral palsy, and autism conditions. The complex involuntary hand movements were felt to be the distinctive feature of the syndrome (Al-Mateen et al., 1986). Leiber (1985) stated that the peculiar stereotyped motions are the leading symptom and are of utmost significance.

From interpreting the existing literature, hand movements may best be categorized as follows: (a) reduction in purposeful use of the hands; (b) absence of purposeful use of the hands; (c) stereotypical hand movements with the hands usually apart; (d) stereotypical hand movements with the hands usually together, and (e) age at which hands were more often in midline position.

Evidence of <u>reduction</u> in purposeful use of the hands was reported in several studies. Naidu, Murphy, and Moser (1986) concluded that hand abnormalities and poor use occurred in all 70 clients between the ages of six months and two years. Witt-Engerstrom (1987) reported that a partial or total loss of acquired manipulative hand skill carried a predictive value and could be observed as early as 15 months in some clients.

Al-Mateen et al. (1986) hypothesized the loss of purposeful hand movements may be a progressive apraxia.

Fontanesi and Haas (1988) also suggested the loss in intentional hand use may be due to a motor dyspraxia.

Some studies found the reduction in hand use preceded stereotyped hand movements, while others found the reverse. Nomura, Segawa, and Higurashi (1985) reported purposeful hand use was lost before the stereotyped hand movements began. Trevathan and Naidu (1988) stated loss of hand skill may be the most reliable early sign, and precedes stereotypic hand movements. Witt-Engerstrom (1987) also found a similar relationship with loss in hand skill preceding specific hand stereotypes. Budden's (1986) data reflected the opposite relationship with stereotyped hand movements usually occurring prior to the loss of hand function. The discrepancy of the Witt-Engerstrom (1987), Trevathan and Naidu (1988), and Nomura et al. (1985) studies with the Budden (1986) study may be one of definition. It is not clear whether "loss" in "loss of purposeful hand movements" was to be interpreted as a reduction or absence of purposeful hand use.

Studies usually reported the most common stereotypic hand movement behaviour as hand-washing, rubbing the hands together, or kneading movements. Olsson and Rett (1987) stated the most common stereotypes are hand-washing movements, pressing the hands together in front of the chest or chin. Kulz et al. (1985) described a single case as incessant kneading, rubbing the hands, and pressing the

hands together. Stereotyped hand movements were described by Percy, Zoghbi, and Riccardi (1985) as the hands clasped together in a wringing type movement. Hanefeld (1985) described stereotyped hand movements as washing and knitting movements.

Though the hand washing movements are usually the predominant stereotypes, other stereotypes were presented. Zapella (1985) described stereotypical hand movements to include hand clapping. Brunel and Gilly (1985) expanded on stereotypical movements describing a client as raising her hands to her mouth, putting them in and out of her mouth, rubbing and wringing them in front of her mouth, and stroking her cheeks. The Naidu et al. (1986) study of 70 cases included the following stereotypical hand movements: twirling the hands by the side or above the head, tapping the chest or chin, pulling of the hair or ears, rubbing the nose, hand to mouth with biting or rubbing of the hand against the lips and teeth. Naidu et al. (1986) reported stereotypical hand movements moved towards midline at 18 months to 2 years. Kerr et al. (1987) gathered information for 40 subjects' early history by interviewing parents, and if films were available of the child's early years, these were analyzed as well. The purpose of the Kerr et al. study was to observe hand movements before, during, and after the regression. Descriptions of hand movements were listed for the periods

prior to, during, and following the regression. It was concluded that before the regression the hands were usually separate, during the regression usually together, and thereafter with increasing age, the teenage years and older, hands tended to separate again.

Hand washing and mouthing appeared to be the most obvious and frequently reported stereotypes; however, other stereotypes such as repetitive clapping, waving, and rubbing hands against the face or chest were reported to occur. Hand movements are important to include in the study of Rett syndrome. Hand movements are cited in the four stage clinical description, the diagnostic criteria, and in numerous investigations of RS. In reviewing the existing literature hand movements could be categorized into five types: (a) reduction in purposeful hand use, (b) absence of purposeful hand use, (c) stereotypical hand movements with the hands usually apart, (d) stereotypical hand movements with the hands usually together, and (e) age when hands were more often in midline position.

Motor Development

To understand the communication development/regression of children with RS it is also necessary to collect information in other areas of development. Previous investigations have provided enlightening information with regard to motor milestones. Hanefeld (1985) described a variety of clinical behaviours and medical symptoms of a

child with Rett syndrome. Hanefeld (1985) included the following information on motor milestones: Sitting, 9 months, pulling self up, 10-12 months, crawling, 12 months, and never able to walk unsupported. Budden's (1986) study provided information on 12 subjects concerning the onset of walking and age at the end of early normal development. Two subjects did not walk and the other 10 subjects were able to walk between 12 and 36 months, with a mean age of 19 months. The end of early normal development ranged from 9 to 30 months, with a mean age of 13 months. Al-Mateen et al. (1986) determined that all 15 subjects experienced a period of normal development ranging from the first four to 18 months of life. Information was provided for the motor milestones of sitting without support and walking independently. All 15 subjects had acquired the ability to sit without support, but only 10 of the 15 girls had walked independently.

Fontanesi and Haas (1988) studied cognitive development using the Vineland Adaptive Scale, the Bayley Scales of Adaptive Ability, and Piaget's Object Permanence subtest to assess 18 RS clients. Three of the 18 subjects were diagnosed as atypical and 15 were diagnosed as typical RS. The age of subjects were 2 1/2 to 23 years. In addition to the cognitive information the investigators gathered information on the month children acquired the

following motor milestones: sit, crawl, stand, and walk. The motor milestone information was obtained through parent interview and if available, medical charts. All clients were able to sit, but as the motor skills became more complex, fewer children acquired them. The mean ages in months were calculated using information in months for the children who had acquired the milestones, and the results were as follows: sit, 6.0 mos.; crawl, 10.4 mos.; stand, 13.1 mos.; and walk, 21.0 mos.

One of the most comprehensive investigations of motor milestones was completed by Nomura et al. (1985). The motor milestone information was correlated with age dependent appearance of certain symptoms and head circumference. Nomura et al. (1985) listed either the "month" or "never acquired" for each of the following milestones: head control, rolling over, sitting when placed, sitting by themselves, crawling, standing with support, standing without support, walking with support, and walking without support. The investigators also noted the characteristic walk involved a broad based stance with short steps and lack of coordinated movements of the upper extremities.

Nomura et al.'s (1985) investigation and other studies commented on an abnormal crawl. All 11 subjects had experienced difficulty with crawling (Nomura et al., 1985). Nomura et al. (1987) studied six cases and

commented that hypotonia is present in infancy, and is noticeable in later infancy when the child experiences difficulty with crawling. When a pair of twins with RS began to crawl they crawled on one knee and pulled themselves along with their arms (Coleman et al., 1987). Twenty percent of 70 subjects did not crawl or had an abnormal crawl, that was sometimes described as a bunny hop, where the legs were used but not the hands (Naidu et al., 1986). Some of the 20% just rolled like a log instead of crawling.

In most cases motor development appeared normal for at least the first few months, but later developmental milestones, such as sitting unassisted or walking, were delayed (Olsson and Rett, 1987).

Decrease in Interactions

The term "decrease in interactions" will include the avoidance of eye-contact, quietness, avoidance of verbal or nonverbal initiations with people, and a decrease in play.

In Gillberg's (1987) investigation eight mothers of RS children, 10 years old or younger, completed a questionnaire that was originally developed to discover early symptoms in infantile autism. When the mothers were asked what was the first worry that alerted them to seek medical help, two of the eight mothers commented about

their child's social interactions or reaction to the environment: regression of social language and motor skills, and failure to imitate in social interaction. Gillberg's (1987) investigation found social behaviors that were similar for both RS children and autistic children. These included the following: overjoy when tickled, lack of social initiatives, marked periodicity, empty gaze, sleep problems, and preoccupations with odd objects. Three statements that were consistently evident for autistic children but rare in RS children were as follows: did not like to be disturbed, played only with hard objects, and very pleased when left completely alone. Gillberg (1987) found that RS children were not as uninterested in social interaction as autistic children. Though the Rett syndrome children did not actively seek human contact they did not mind human interactions or seem disturbed by them.

Kerr et al. (1987) studied 40 RS children and reported that during the period of regression the children appeared confused and withdrawn. Coleman et al.'s (1987) case history information described RS twins who gradually became withdrawn, began to lose eye contact, and lost their joyous expressions. Olsson and Rett (1987) reported poor interactions in social situations, smiling or laughing without apparent reason, extreme quietness, long daytime sleep, lack of interest in the environment, and

lack of eye-contact. Olsson and Rett (1987) observed that most autistic children actively avoided eye-contact, while RS children smiled at faces, looking into their eyes; however, they did not accommodate to faces or fix on faces. This behaviour gave observers the impression that the RS children were looking through the person (Olsson, 1987; Olsson & Rett, 1987). The Rett syndrome children seemed happier when people were near. Olsson and Rett reported the Rett syndrome children smiled at unfamiliar people in an unfamiliar room, with the same frequency as they smiled at their parents. This was interpreted by Olsson and Rett as highly retarded social and affective development (Olsson, 1987; Olsson & Rett, 1987). Olsson (1987) described Rett syndrome behaviours which were similar to symptoms of severe dementia such as: a restricted repertoire of mental and motor performances, monotonous in speed and form; a small number of stereotypic movements, monotonous in speed and form; weak or insufficient responsiveness to the social environment; and amimia, constant facial expression.

Witt-Engerstrom's (1987) investigation of 10 Rett syndrome children presented information on social contact. Information included the following: at 9-10 months emotional contact was essentially normal; at 15 months emotional contact was impaired in four of the girls, diminished interest and contact were reported; at 24

months only three children gave some contact and emotional response. Partial or total loss of interest/play and contact were concluded to be some of the predictive variables of stage II Rett syndrome. Evidently, the type of decreased interactions differ in autism and RS and may occur in RS as early as 15 months.

Receptive and Expressive Language

Researchers have alluded to vague or general statements of communication development and regression rather than focusing on details of communication milestones and regression for the birth to 24 month stage. A few studies included one to three communication milestones as variables, but investigations were not found that studied communication development\regression in further detail. General statements of communication abilities are listed in the diagnostic criterion, the four stage clinical description, and in case histories of some investigations.

The diagnostic criterion (Appendix A) includes a general statement of communication abilities: severely impaired expressive and receptive language (Trevathan & Naidu 1988). The four stage clinical description (Appendix B) also includes general statements concerning communication as follows: Stage I, Changed communicability; and Stage II, Severe dementia (Hagberg &

Witt-Engerstrom, 1986; Trevathan & Naidu, 1988). A broad statement about communication may be adequate for the purpose of inclusion in the diagnostic criterion, because communication development is variable and the inclusion of further information may create an artificially narrow or strict criteria. However elaboration on communication development for the RS description would be valuable. Details of communication development/regression within the clinical description may better prepare professionals to monitor the child's development and progression of the syndrome, and allow them to provide appropriate counselling to parents regarding expectations and goals.

Rett syndrome medical studies have concentrated on medical symptoms or variables but have occasionally included one or two communication variables. Goutieres and Aicardi's (1986) metabolic investigation of 12 RS females included the statement that all girls had "lost" or "never acquired language". Al-Mateen et al. (1986) listed whether the 15 clients had "lost" or "never acquired speech" (9-lost, 6-never acquired). Another medical study graphed the percentage of cases that evidenced poverty of babbling in infancy (approx. 70%), and mentioned the loss of verbal expression and speech delay after the age of one year (Nomura et al., 1985, p.336). Kerr et al. (1987) studied the subjects hands pre- and post-regression, and included the "number of

words acquired" (range of 2 to 10 words), and if the 20 girls had ever used "two word phrases" (2 girls used one two word phrase).

Investigations reporting case studies or behavioral observations usually mentioned a general statement of communication abilities when reporting the clinical history. Hanefeld's (1985) clinical history of one child with RS included the statement that intelligible speech was not acquired. Tariverdian et al. (1987) described twins who had become severely mentally handicapped, as able to babble but having no language. In Kulz et al.'s (1985) single case study, single words were evident at 15 months, speech regressed at 18 to 20 months, and at 3yrs-8months the child was unable to understand speech or express herself with speech. Witt-Engerstrom's (1987) investigation of early predictive symptomatology included a few comments about communication: at stage II communication abilities became poor and at stage III girls whispered some partial words and started babbling again.

Studies comparing autism and Rett syndrome provided information regarding communication, but again the statements were general. These studies investigated characteristics of the stage where Rett syndrome and autistic children shared similar behaviours, and as a result, examined children in the preschool or school-age years rather than the first few years. The reason

detailed descriptions of communication were not provided in studies comparing infantile autism and RS is because communication variables were not found to be a distinguishing feature. The diagnostic criterion of infantile autism included onset prior to age three, and severe speech anomalies or no speech at all (Olsson, 1987). This definition of infantile autism would also describe RS. Some of the most striking differences between autism and Rett syndrome are as follows: autistic children follow a non-progressive course (Gillberg, Walhstrom, & Hagberg, 1985), demonstrate better gross and fine motor abilities and better use of the hands, and make an obvious effort to avoid eye-contact (Gillberg 1986, 1987; Olsson, 1987; Olsson & Rett, 1987).

Zapella's (1985) study of autism and RS described the typical pattern of communication for 20 children with RS as normal for the first year or few years. Some of the children began to say a few words and even a few sentences in the second year. Zapella (1985) also provided a more detailed description for 3 of the 20 children. Descriptions of verbal abilities from Zapella's (1985) case history reports were as follows:

Case One:

> At one and a half years this girl began to utter her first words. Her vocabulary increased subsequently and she began to use short phrases.

Case Two:

Between 9 months and 1yr-3months this child's vocalizations reduced, at 1yr-3months she was already unable to utter a sound, and presently is unable to say words.

Case Three:

This child's vocalizations were adequate, and at one year she started to say her first words. Her vocabulary only increased to 15-20 words, and at 2yrs-4months she lost most of her ability to speak.

One of the most detailed descriptions of communication abilities was found in a study of 70 RS children by Naidu et al. (1986). This study described numerous medical and behaviour symptoms and listed verbal abilities, but not receptive language abilities. The information was compiled and described for the ages 0-6months, 6-12months, 12-18months, 18months-2yrs, 2-5yrs, and 5-15yrs, and greater than 15 years. Descriptions of verbal abilities were extracted and are listed as follows:

0-6months: No statement of communication ability.

6-12months: Children produced the words "mama" and "dada" or said two or three words. Lack of progression of language skills was noted.

12-18months: New words were not acquired, and the words previously acquired were not used frequently.

18months-2yrs: Severe language arrest.

2-5yrs: No statement of communication ability.

5-15yrs: Language was either totally absent or remained at two or three words or phrases. Two nonverbal children uttered a meaningful sentence when febrile or stressed.

> than 15yrs: Language was either absent or a few words were used. If children used a few words, they were usually inappropriate for the situation.

From the case studies it may be concluded that there is some variability in communication development from birth to 24 months, but children with RS typically become nonverbal and evidence a significant decrease in communication abilities between the ages of one and three and a half years.

Previous investigations of RS have provided very limited information on early linguistic development, and no information on preverbal or prelinguistic development. A major marker for prelinguistic development is the emergence of intentional communication (Wetherby, Cain,

Yonclas, & Walker, 1988). Children's communicative intentions are expressed first through the use of gestures and then verbally (Bates, 1979; Coggins & Carpenter, 1981; Masur, 1983; Zinobar & Martlew, 1985). The types of gestures most commonly investigated in communication research are showing, giving, and communicative pointing (Bates, 1979; Coggins & Carpenter, 1981; Leung & Rheingold, 1981; Masur, 1983; McLean & Snyder-McLean, 1987; Volterra & Caselli, 1985; Zinobar & Martlew, 1985). These investigations support an age range of 9-13 months as the stage where communicative gestures such as pointing, showing, and giving are usually present. Data on the emergence of communicative gestures would indicate if and when the RS child achieves these prelinguistic communicative behaviours.

In summary, research on RS children has not focused on linguistic and prelinguistic communication development in a comprehensive manner. A few studies included one or two communication milestones, but there were no studies that investigated the sequence of communication milestones from birth through to 24 months. Such an investigation could provide a more comprehensive picture of communication development in the early years of a RS child's life.

Area of study

Researchers have commented on the need for further research of the Rett syndrome. Opitz (1966) stated that it is collaborative work with Rett syndrome children, their families, and our fellow scientists that will ultimately decipher the mystery of Rett syndrome. Rolando (1985) reported that an increased widespread knowledge of Rett syndrome is necessary to reduce the troublesome and time-consuming search; to reduce the number of investigations to which patients are usually submitted; to relieve parents of guilt; and to provide realistic and clear suggestions for adaptive and therapeutic programs.

The purpose of this investigation was to address the gap in existing knowledge on early communication development in children with RS. The study explored prelinguistic and early linguistic development and regression.

Based on existing information on communication development and information reported in other developmental domains, it was expected that RS children develop normally initially, followed by a slowing of development, and finally a regression, losing previously acquired abilities. In this investigation a communication questionnaire was mailed to parents' of two to four year old RS children. It was then possible to determine at what point RS children generally began to fall behind the

typical communication development sequence. In addition, by having parents report on their child's current abilities it was possible to describe the extent of the regression of previously acquired communication milestones.

Retrospective Study

Previous investigations of RS have relied on retrospective data collection procedures (Gillberg et al., 1987; Kerr et al., 1987; Witt-Engerstrom, 1987). Witt Engerstrom (1987) utilized retrospective data collection procedures to study early predictive symptomatology. The motivation for the investigation reportedly arose from the frustration expressed by parents who after reporting their child's early behavior changes to health professionals, were met with assurance that their child was normal (Witt-Engerstrom, 1987). Witt-Engerstrom (1987) stated if the early and quite peculiar deviations could be distinguished, this information would be valuable for early detection and prediction. Witt-Engerstrom (1987) asked parents of 10 RS children, aged 20 months to 6 1\4 years, questions based on the Denver Developmental Screening Test, to recall information about their child's development at 6, 9-10, 15, 18, and 24 months. Conclusions were reported in a descriptive manner.

Most investigations have relied on retrospective parent report for detailed case histories. Kerr et al.

(1987) reported that retrospective developmental histories have obvious weaknesses, but in their investigation the encouragement to recall specific details of behavior produced precise and useful information. Kerr et al. (1987) relied on retrospective developmental histories to collect information on hand movements. Gillberg et al. (1987) utilized a questionnaire to gather data from parents of 8 children with RS, to investigate differences between autism and RS. Responses to 130 questions involved having mothers indicate with an X on a line 100 mm long, whether a statement "did not" or "did" apply. Examples of statements included in the questionnaire are as follows: "did not seem to react adequately to cold", "did not like other children", and "strange reactions to sounds".

Test results from the first few years rarely exist; consequently, health professionals generally need to rely on retrospective parent report for the RS child's history. Philippart (1986) reported that consultants first see the children several months or years after the regression has commenced. A Swedish study cited the mean age of referral for RS children was 3 1/4 years (Witt-Engerstrom, 1987). The late diagnosis precludes the possibility of a prospective research study when investigating the first few years of development. One method of collecting information retrospectively is by parents' completion of

a questionnaire. A questionnaire allowed the investigator to reach an adequate number of children, and to collect information that was not possible to collect prospectively due to the late diagnosis.

Questionnaire

The questionnaire in this investigation was based largely on the Clinical Linguistic Auditory Milestone Scale (Capute, Palmer, Shapiro, Wachtel, Schmidt, & Ross, 1986), which focuses on milestones from birth to 24 months. There were two limitations to the CLAMS for the purposes of this investigation. First, although it was not expected that RS children would develop communication milestones past 24 months, additional items past 24 months were required to ensure the questionnaire covered the highest level of development for all RS children. Specific communication acquisition reported by Miller (1981) from 29 months to 45 months was added. The second limitation of the CLAMS was that it did not go into extensive detail on intentional communication for the prelinguistic stage of language development. The questionnaire was augmented with gesture questions focusing on the giving, showing, and pointing gestures reported in the literature. Finally, additional items on hand movements were added to the questionnaire to determine if hand movements were an interfering factor in

the development of gestures. Questions were based on hand movements reported in past investigations of RS.

Questionnaire: Clinical Linguistic and Auditory Milestone Scale

Capute et al. (1986) developed the milestone scale based on existing normative data. They then conducted an investigation to gather normative data on each of the 25 milestones from their scale, and to determine if the CLAMS reflected differences between a normative group and a delayed group. The normative subjects were 198 males and 183 females from the total population who were born at full term. They had normal neurological examinations during the second year of life, and normal Bayley Mental (MDI) and Psychomotor Development indices (PDI) at one year of age (Capute & Accardo, 1986). The population consisted of predominantly white subjects from all social classes, but with a large number from the middle class. The delayed group comprised of 15 infants that were greater than one standard deviation below the mean on the Bayley Scales of Infant Development. The total population of 'average', 'delayed', and premature subjects was 448.

The subjects were followed prospectively from birth to age 24 months. Assessment visits were scheduled at the child's age of 2 weeks, 2, 4, 6, 9, 12, 15, 18, and 24 months. Parents were asked to report on milestones at each visit. The children were then assessed by a pediatrician

to determine milestones attained. In addition all infants were assessed at age one by a psychologist who was unaware of the milestone data using the Bayley Scales of Infant Development.

The 'delayed' subjects attained each milestone at a later age than the 'average' or normative subjects. In addition, the correlation between the BSID scores and milestone performance on the CLAMS was statistically significant.

The data for the normative population (Appendix C) indicated that milestones are attained in an orderly, sequential fashion with a narrow range of variability for each milestone after the first few weeks of life. It was also reported that parental recall for the milestones was above 80% for 13 of the 25 milestones and above 70% for 21. The normative data included the mean age of attainment and the standard deviation for each receptive and expressive language milestone.

Questionnaire: Intentional Communicative Gestures

The most commonly investigated communicative gestures of giving, showing, and pointing have been reported to emerge during the 9 to 13 month stage. Definitions for these gestures have been reported by Bates (1979 p. 132).

Questionnaire: Atypical Hand Movements.

The gestures reflect the children's first signs of intentional communication in the prelinguistic stage of language development. If early communicative behaviors such as gestures were absent it had to be determined if this was thought to be due to a delay in communication development or a decreased ability to use the hands/arms for functional purposes (grasping, mobility). Questions regarding hand mobility/usage were asked to determine if a decrease in hand control was or was not a confounding variable in interpreting questions related to use of gestures. The questionnaire asked questions on hand movements based on information reported in the literature.

A review of hand movements, motor development, involvement in interactions, and language development enables one to begin to perceive the early development of children with Rett Syndrome.

# CHAPTER 3

## Method

This investigation was a survey study (Jackson, 1988). The information is presented in a descriptive manner.

### Subjects

Subject participation, parent(s)/guardian(s) of Rett syndrome children, was voluntary. Each subject's diagnosis was reported to have been confirmed by a physician. One question on the questionnaire addressed whether the diagnosis was reported to be definite or uncertain. If the diagnosis was uncertain, the data from that questionnaire were excluded. Researchers have determined that a child must meet all of the eight criteria (Appendix A) before being diagnosed as typical or classical RS (Goutieres & Aicardi, 1986, 1987; Hagberg & Rasmussen, 1986; Hagberg & Witt-Engerstrom, 1986; Percy et al., 1987; Zapella, 1985). The focus of this study was on subjects that had a definite RS diagnosis.

All children were between the ages of 23 to 48 months. Age 23 months was chosen as the lower limit because the diagnostic criterion for RS is often tentative until at least age two years (Trevathan & Naidu, 1988), and because the mean age of referral for RS children was reported to

be 3 1/4 years (Witt-Engerstrom, 1987). Age 4 years was chosen as an upper limit in order to offset the effects of inaccurate recall of the child's early developmental period. While there is no data available to guide in the selection of an upper limit, the chosen age limit was more stringent than in two other retrospective studies (ie. Kerr, et al. 1987; Witt-Engerstrom, 1987).

Subjects were the 17 parents who completed the questionnaires. The questionnaires were sent to parents of the 40 youngest children registered with the International Rett Syndrome Association. The survey return rate was 55%. Eighteen subjects did not return the questionnaire; twenty-two subjects did return the questionnaire. Five did not meet the subject selection criteria; one of the five was misdiagnosed with Rett syndrome and later diagnosed with Angelman syndrome, one was male and thought to have had cerebral palsy, and three did not have a confirmed diagnosis. The subjects that met the subject selection criteria consisted of parents of 17 RS children that were between the ages of 23 to 48 months with a mean of 39 months. Each child's age is reported in Appendix M. Fabricated initials were assigned to each subject's child to assure anonymity.

Procedure For Pilot Study

A pilot study was undertaken to assist in developing the questionnaire. A first phase was to determine if any directions or items on the questionnaire were ambiguous or unclear. The questionnaire was given to three parents of normal children that were between the ages of 2 1/2 and 5 1/2 years. Parents known to this investigator read the questionnaire and answered the questions. They were asked to comment on anything that was unclear. Feedback was provided to the principal investigator and a few sentences were revised on the questionnaire.

A second phase of the pilot study involved sending questionnaires to a few speech-language pathologists to give to parents of children between age 2 1/2 and 6 1/2 with a documented language delay of moderate of greater severity. A total of 18 questionnaires were sent out with a return rate of 61%. A second copy of the questionnaire was sent out to determine parents' reliability across two completions of the questionnaire. Seven of the 11 respondents completed the second questionnaire. A 70% criterion for reliability was chosen based on the fact that the authors of the CLAMS reported prospective parental recall for the milestones was above 80% for 13 of the 25 milestones and above 70% for 21. It was expected that retrospective parental recall would not be as accurate as prospective recall; however a reasonably high

percentage of 70% was chosen. Reliability across two completions of the questionnaire was sufficient for checking observed or not observed (x=84%) for a milestone but not sufficiently reliable (x=58%) for circling an age estimate (A vs B vs. C). When the choices were collapsed into "normal" (A) versus delayed (B or C) reliability was sufficiently high (x=78%).

It was impossible to run the pilot study on parents of young Rett syndrome children and as a result both types of responses to the questions (checking observed/not observed and circling an age estimate) remained in the questionnaire as the circled age estimate data could be deleted or collapsed into normal and delayed after the fact if reliability was not sufficiently high.

Procedures

The Canadian Rett Syndrome Association were provided with packages that included the questionnaire (Appendix D), cover letter (Appendix E), consent form (Appendix F), and check form (Appendix G). They mailed the packages to subscribers of the Canadian Rett Syndrome Association Newsletter who had indicated that they were the parents of a child with RS. The Association did not have information on the age of the children. Participation was voluntary. If one chose to participate, the consent form was signed, and the completed questionnaire was returned. The

parent(s)/guardian(s) retained a copy of the consent form. If recipients chose not to participate they were requested to return the questionnaire.

Data collection occurred during a six month period. None of the returned questionnaires were within the subject selection age range.

The research proposal was then sent to the International Rett Syndrome Association in the United States. The Association had a data base which allowed them to determine the current age of the children with Rett Syndrome. As a result questionnaires could then be mailed specifically to the parents of the forty youngest RS children within the subject selection age range. The International Rett Syndrome Association mailed 40 questionnaires to families of the 40 youngest RS children in the Association's data base that were within the age range. The packages included the cover letter (Appendix H), the questionnaire (Appendix D), two copies of the consent form (Appendix I), and the check form (Appendix J). Data collection occurred during a six month period.

Questionnaire (Appendix D)

The questionnaire consisted of the following five areas:

PART A: Questions focusing on demographic data and diagnosis.

PART B: Questions based on the Clinical Linguistic and Auditory Milestone Scale (Capute et al., 1986) examining receptive and expressive milestones from birth to 24 months. Questions focusing on milestones from 19 to 46 months were added to ensure a ceiling and have been based on developmental language data reported by Miller (1981).

PART C: Questions on prelinguistic intentional communication were based on the three primary gestures (pointing, showing, giving) reported in the literature.

PART D: Questions examining the child's fine and gross motor control, and atypical movements. This was necessary to determine if there are any possible interfering relationships between the onset of gestures and atypical hand movements. The questions were based on information on hand movements reported in previous RS studies.

PART E. Questions focusing on the child's present level of functioning based on milestones from the CLAMS (Capute et al. 1986). Parents were requested to place a checkmark by the behaviors/milestones that reflected what their child was currently demonstrating. The information allowed for comparisons with part A (highest level of development) with part E (current level of development) to determine the extent of regression for each RS child.

## Reliability

### Intrajudge and Interjudge Reliability:

All results were tabulated by the principal investigator. Two sets (12%) of the questionnaires were randomly selected for intrajudge reliability in tabulating the questionnaire results. The point to point agreement was 100%. A second judge, a speech-language pathologist, was requested to tabulate the results for a second random selection of 12% of the questionnaires for interjudge reliability. Again the point to point reliability was 100%. A high agreement was anticipated as tabulating the results was a straightforward task.

### Subjects' Reliability Across Two Completions of the Questionnaire:

A second copy of the questionnaire was mailed to the parents who had completed the first copy of the questionnaire and had agreed to participate. This procedure was undertaken to determine the reliability of parent report on the questionnaire questions.

A cover letter (Appendix K) and second copy of the questionnaire were mailed out to the 17 subjects three weeks after they had returned the initial questionnaire. Ten subjects returned a second completed copy of the questionnaire. The 12 sets of questionnaires were submitted to an item by item analysis. An agreement was

defined as checking/circling the same answer on both questionnaires. Determining the reliability was necessary to ascertain whether the data should be reported as observed/not observed, or whether parent report was sufficiently reliable to report the degree of delay (circled responses A,B,C).

CLAMS QUESTIONS: The reliability results revealed that parents were consistent or reliable (89%) in their responses to questions about whether milestones or the CLAMS questions **were or were not observed** (>than 70%). They were not reliable at circling a specific age estimate (A vs. B vs. C) for when milestones were observed. When the data was collapsed into **normal versus delayed** (A vs. B or C) reliability was just beyond 70% but judged as not sufficiently high because only four parents were above the reliability criterion. The reliability results for the 25 CLAMS' questions are presented as follows in table 1.

TABLE 1 **RELIABILITY-CLAMS** N=12
**Agreements**

| Subjects: | Observed/not observed: | Degree of delay Circling A,or B, or C: | Delayed or not delayed. Circling A vs. B or C: |
|---|---|---|---|
| R.T. | 21/25=84% | 5/11=45% | 7/11=64% |
| L.E. | 22/25=88% | 7/11=64% | 7/11=64% |
| O.N. | 24/25=96% | 3/7=43% | 6/7=86% |
| A.N. | 21/25=84% | 9/17=56% | 11/17=65% |
| R.Y. | 23/25=92% | 4/8=50% | 5/8=63% |
| N.O. | 25/25=100% | 7/7=100% | 7/7=100% |
| C.K. | 25/25=100% | 6/12=50% | 7/12=58% |
| I.N. | 21/25=84% | 8/8=100% | 8/8=100% |
| N.N. | 23/25=84% | 6/13=46% | 8/13=62% |
| A.M. | 20/25=80% | 10/16=63% | 10/16=63% |
| G.E. | 23/25=92% | 8/15=53% | 9/15=60% |
| E.R. | 21/25=84% | 5/5=100% | 5/5=100% |
| | Mean $\bar{x}$=89% | $\bar{x}$=64.17% | $\bar{x}$=73.75% |
| | S.D.=6.8 | S.D.=22.55 | S.D.=17.29 |
| | Range=80%-100% | Range=43%-100% | Range=58%-100% |

Parent reliability for reporting whether a milestone was or was not observed with a mean of 89% <u>was</u> sufficiently high (each score above 70 percent). All subjects were above the 70% criterion. Reliability for reporting when a milestone occurred by circling A, B, or C <u>was not</u> sufficiently reliable with a mean of 64.17% for selecting A, or B, or C. When the data were collapsed into a within normal (A) or a delayed by 2 or more S.D.s (B or C) the reliability <u>was not</u> sufficiently reliable at 73.75% because only 4 of the 12 subjects were above the

70% criterion. Consequently parents' responses for selecting observed or not observed for the CLAMS milestone questions (Questions 1 to 25) will be reported in the data analysis section.

Individual milestone questions were evaluated to determine if reliability for circling A, B, or C was sufficient (70%) for some of the milestones. If less than 3 of the 12 subjects checked observed for a milestone then that milestone was excluded from the analysis. The number of agreements for circling age categories A, B, or C were recorded across all subjects for each milestone question. Responses for circling an age category on the following milestone questions were found to be sufficiently reliable.

TABLE 2: **RELIABLE MILESTONE QUESTIONS** N=12

| Milestones: | Agreements | % |
|---|---|---|
| Question 1: Receptive: Alert | 9/11 (11 subjects chose observed, 9 responded the same across two completions) | 82% |
| Question 2: Receptive: First Smiled | 8/11 | 73% |
| Question 6: Expressive: Raspberry Sound | 6/7 | 86% |
| Question 9: Expressive: Said "dada" and "mama" indiscriminantly | 7/10 | 70% |
| Question 10: Expressive: Said "mama" referring to mother | 5/7 | 71% |
| Question 11: Expressive: Said "dada" referring to father | 5/7 | 71% |
| Question 13: Expressive: Two words other than mama and dada | 5/6 | 83% |

One must take caution not to over-interpret the above data and conclude that expressive milestones were much more reliable than receptive milestones, because some expressive milestones received quite low reliability scores. Excluding the milestones where less than three subjects had checked "observed", the reliability for all of the receptive milestone questions was 53% and the reliability for all of the expressive milestone questions was 61%.

Subjects were more reliable on the milestones in Table

2 than other CLAMS' milestones on the questionnaire. It may be that these milestones were more obvious or memorable for these parents enabling them to reliably estimate a time frame in terms of normal (A), delayed by 2 S.D.(B), or delayed by 3 or more S.D.s(C).

OTHER QUESTIONS: All of the parents reported that the Miller milestones questions were not observed on both questionnaires.

Parent consistency or reliability across two completions of the questionnaire for the remaining sections of the questionnaire were as follows.

TABLE 3 **RELIABILITY-REMAINING QUESTIONS** N=12

| Subjects | Gesture Questions Part C (Ques. 1-3): **Agreements** (A=Before 13 mons. B=After 13 months) Obs/Not: A/B: | Hand Movement Questions Part D (Ques 1-6): **Agreements** Obs/Not: A/B: | Other Delays Part D (Ques A-E) Yes/No | Milestones At Present Part E: Obs/ Not obs: |
|---|---|---|---|---|
| R.T. | 3/3=100% 0/0 (0/0 none observed so A or B were not circled) | 6/6= 100% 6/6= 100% | 4/5= 80% | 22/25= 88% |
| L.E. | 3/3=100% 0/0 | 5/6=83% 4/5=80% | 5/5= 100% | 21/25= 84% |
| O.N. | 3/3=100% 0/0 | 3/6=50% 2/4=50% | 3/5= 60% | 23/25= 92% |
| A.N. | 2/3=67% 0/2 | 6/6=100% 5/5=100% | 5/5= 100% | 19/25= 76% |
| R.Y. | 3/3=100% 0/0 | 6/6=100% 4/6=67% | 5/5= 100% | 23/25= 92% |
| N.O. | 1/3=100% 1/1 | 6/6=100% 4/4=100% | 5/5= 100% | 24/24= 96% |
| C.K. | 1/3=33% 0/0 | 5/6=83% 3/6=50% | 5/5= 100% | 20/25= 80% |
| I.H. | 3/3=100% 0/0 | 6/6=100% 6/6=100% | 5/5= 100% | 23/25= 92% |
| N.N. | 3/3=100% 0/0 | 6/6=100% 6/6=100% | 3/5= 60% | 22/25= 88% |
| A.M. | 1/3=33% 0/1 | 5/6=83% 4/4=100% | 5/5= 100% | 23/25= 92% |
| G.E. | 3/3=100% 3/3 | 4/6=67% 2/4=50% | 5/5= 100% | 22/25= 88% |
| E.R. | 3/3=100% 0/0 | 5/6=83% 5/5=100% | 4/5= 80% | 21/25= 84% |
| Mean | x=86.08% | x= 87.42% | x=83.08% | x=90% | x=87.67% |
| S.D. | =28.4 | 26.53 | 16.08 | 15.95 | 5.77 |
| Range | =33-100% | 50-100% | 50-100% | 60-100% | 76-96% |

The mean reliability for reporting whether gestures were observed or not observed was sufficiently reliable at 86.08%. Most gestures were not observed; consequently determining whether they were delayed or not delayed was not possible. In the data analysis section gestures will be reported as observed or not observed.

The mean reliability for reporting whether hand movements were observed or not observed was reliable at 87.42%. In addition parents were reliable (83.08%) at indicating whether these hand behaviors occurred before or after 13 months.

Parents were reliable (90%) in reporting "yes" or "no" fcr noticing other developmental delays (ie. gross motor, fine motor etc.). Parents were then requested to estimate the age of their child when they had noticed each delay. An agreement was defined as reporting an age estimate within 4 months +or- across two completions of the questionnaire. The reliability was 79.75%.

Section E of the Questionnaire involved parents checking off which milestones (CLAMS) their child presently demonstrates. The reliability was sufficiently high at 87.67%.

The reliability analysis was necessary to determine how the data should be reported in the data analysis section. The criterion for reporting the data was set at a mean reliability score of at least 70%.

# CHAPTER 4

## Results

The results from the questionnaire are presented in a descriptive format based on methods reported in other observational and survey studies on RS children (ie. Fontanesi & Haas, 1988; Kerr et al., 1987; Witt-Engerstrom, 1987).

The biographical information is reported in Appendix L.

The purpose of this investigation was to explore prelinguistic and early linguistic development and regression in children with Rett syndrome. The results from the questionnaire are organized to answer the following three research questions.

1. **Which communication milestones that occur in the stage of birth to 24 months are achieved by children with RS?**

2. **What is the extent of the regression of communication milestones in RS children presently between ages 23 and 48 months?**

3. **When are other developmental delays observed?**

## Experimental Question 1: Communication Milestones Achieved and When.

I. CLAMS Milestones (part B questions 1-25):

Items from the CLAMS questions involved the respondents answering whether a milestone was or was not observed and then circling an age estimate. The mean reliability of 89% for reporting whether a milestone was or was not observed was sufficiently high (criterion 70%) to describe the results. The results are illustrated previously in Table 1. This includes the percentage of children that did not experience the milestone or behavior at some age, and the percentage that did experience the milestone. The results for the CLAMS milestones are in Table 4 as follows.

Table 4 **CLAMS MILESTONES** N=17 subjects

| CLAMS Milestones: | Percent of children that attained the milestone: |
|---|---|
| Alert | 88% |
| Smile | 94% |
| Coo | 82% |
| Orient | 82% |
| Ag-goo | 53% |
| Razz | 59% |
| Babble | 94% |
| Mama/dada indiscriminately | 82% |
| Gesture (wave bye) | 35% |
| Dada discriminantly | 47% |
| Mama discriminantly | 53% |
| One step command with gesture | 18% |
| First word | 53% |
| Immature jargon | 35% |
| Second word | 35% |
| Three single words | 24% |
| One step command without gesture | 18% |
| Four to six single words | 6% |
| Mature jargon | 12% |
| Five body parts | 0% |
| Seven to 20 words | 0% |
| Eight body parts | 0% |
| Two word combinations | 0% |
| Two word sentences | 0% |
| 50 words | 0% |

In viewing table 4 it appears that the overall sample displayed a gradual decline in milestones. A delay became apparent quite early. After the fourth listed milestone, the percentage attained dropped from 82% to considerably lower levels. None of the children attained milestones that were listed beyond the mature jargon level.

Data on the Individual Milestone Questions that were Reliable:

Data on the seven CLAMS' milestone questions that received the highest reliability scores for circling an age estimate (refer earlier to table 2) are listed in the following table.

TABLE 5: **DATA ON THE RELIABLE MILESTONE QUESTIONS** N=17

| Milestones: | Attained within Normal; Choice A: | Delayed within 2S.D.s; choice B: | Delayed by 3 or more S.D.s; Choice C: |
|---|---|---|---|
| Question 1: Receptive: Alert | 10 Subjects | 4 | 1 |
| Question 2: Receptive: First Smiled | 9 | 3 | 4 |
| Question 6: Expressive: Raspberry Sound | 0 | 2 | 8 |
| Question 9: Expressive: Said "dada" and "mama" indiscriminantly | 6 | 5 | 3 |
| Question 10: Expressive: Said "mama" referring to mother | 4 | 2 | 3 |
| Question 11: Expressive: Said "dada" referring to father | 5 | 1 | 2 |
| Question 13: Expressive: Two words other than mama and dada | 1 | 3 | 2 |

The above indicates that some children were delayed from the onset and that many early milestones that occur prior to age two were not attained.

Individual Analysis For Communication Development:

The data were analyzed to determine if subjects' communication delay appeared to cluster into specific categories. Appendix C lists the age norms for each milestone to allow for an estimate of the age level for the highest milestone level. The most predominant pattern was scattered and delayed development to a highest attained milestone level of approximately 9 to 16 months. The majority of children did not surpass an approximate 13 month level. A few children were delayed from the onset and a few others experienced an early abrupt halt in communication development. The patterns are categorized and described in Table 6.

TABLE 6 **INDIVIDUAL ANALYSIS** N=17

| Category Description | Subjects: |
|---|---|
| EARLY AND ABRUPT DELAY:<br>Delayed from first or second milestone and attained communication development to an approximate level of 2 to 6 months. | 1. Y.D.<br>2. E.R. |
| DELAYED FROM ONSET:<br>All milestones delayed or not attained but highest milestone attained reflected an approximate 13 to 16 month communication development level. | 1. I.N.<br>2. N.E. |
| SCATTERED AND DELAYED DEVELOPMENT TO 9-16 MONTHS:<br>A few milestones on target, some milestones not observed or were delayed. Highest communication milestone attained reflected an approximate 9 to 16 month level. Most did not exceed an approximate 13 month level. | 1. T.N.<br>2. N.S.<br>3. N.O.<br>4. L.E.<br>5. O.N.<br>6. B.B<br>7. R.Y.<br>8. C.K.<br>9. A.N.<br>10. N.N.<br>11. R.T.<br>12. G.E.<br>13. A.M. |

II.Later Milestones:

(Part B Questions 26-29, and part C questions)

The later milestone questions adopted from Miller (1981), were evaluated to determine if children with RS fell within the expected age range.

The milestones reported in Miller (1981) added to this questionnaire were as follows.

| Milestone: | Predicted Age: |
|---|---|
| 1. Uses 'what' questions (eg. what is that? Where is it?). | 19-28 months +or- 1SD = 16-34 mons. |
| 2. Use of the negative can't or don't in a sentence (eg. He can't play.). | 20-34 mons. +or- 1SD = 24-41 mons. |
| 3. Auxiliary is inverted and includes modals, "can", "will" or "do" (eg. Will he bring it home?). | 35-38mons. +or- 1SD = 28-45 mons. |
| 4. Uses "why" questions (eg. Why is he running?). | 35-42 mons. +or- 1SD = 28-45 mons. |

The Miller milestones were added to the questionnaire to ensure a ceiling on communication milestone development. These extra upper limit milestone questions were not needed as all children reached a ceiling prior to these questions. None of the children attained these milestones.

III. GESTURES/INTENTIONAL COMMUNICATION Part C:

The principle gestures investigated in the literature are communicative giving, showing, and pointing. The available data indicates that these gestures are evident between 9 and 13 months (Bates, 1979; Leung & Rheingold, 1981; Volterra & Caselli, 1985). Based on the questionnaire results it was determined what percentage of RS children attained these gestures at some point in time. See Table 7.

Table 7 COMMUNICATIVE GESTURES N=17
(Give/Show/Point)

| | Attained no gestures: | Attained 1 of 3 gestures | Attained 2 of 3 gestures | Attained 3 of 3 gestures |
|---|---|---|---|---|
| N=17 | 12/17=71% (12 children did not attain any gestures) | 2/17=12% | 2/17=12% | 1/17=6% |

The communicative gestures of giving/showing/pointing that occur between the ages of 9 and 13 months were usually absent in children with RS. Only 6% of the children attained all three communicative gestures, and 12% attained one or two gestures. The majority of RS children (71%) did not attain any of the three gestures.

IV. Interfering Hand Movements Part D:

Subjects were requested to place a checkmark whether an atypical hand behavior was observed or not, and if observed, circle whether it was observed before or after 13 months. The results are listed in Table 8.

TABLE 8 **ATYPICAL HAND BEHAVIORS** N=17

| Atypical hand behaviors: | Observed: | Not Observed: | Before 13 Mons: | After 13 Mons: |
|---|---|---|---|---|
| 1. Noticed a decrease or reduction in purposeful use of the hands (eg. less grasping or holding onto toys, less ability to manipulate objects such as a spoon or toy). | 94% | 6% | 5/16= 31% | 11/16= 69% |
| 2. Noticed a point in time when purposeful use of the hands was totally absent (ie. unable to grasp or hold onto objects). | 82% | 18% | 2/14= 14% | 12/14= 86% |
| 3. Noticed hand(s) repetitively pulling or tapping on other parts of the body (ears, chest, or mouth, etc.). | 82% | 18% | 2/14= 14% | 12/14= 86% |
| 4. Noticed hand(s) repetitively squeezing into a fist and then releasing. | 82% | 18% | 2/14= 14% | 12/14= 86% |
| 5. Noticed hand(s) waving at sides of the body or in the air. | 53% | 47% | 2/9= 22% | 7/9= 78% |
| 6. Noticed hand wringing behavior. | 82% | 18% | 2/14= 14% | 12/14= 86% |

There was a high incidence of the all of the atypical hand behaviors (82-94%) with the exception of hand waving (53%). Most subjects checked observed for four to six of the above atypical hand behaviors. The atypical hand behaviors generally occurred after 13 months (69-86%).

An additional question was provided that asked the parents of the children that did evidence gestures to circle if gestures had occurred before atypical hand movements, if atypical hand movements occurred before gestures, or if they occurred about the same time.

The six hand movement questions and the additional temporal relationship question were analyzed to discover whether a potentially confounding relationship existed between atypical hand movements and the development of gestures. The onset of gestures and the onset of interfering hand movements were compared to determine whether the atypical hand movements occurred before, during, or after the development of gestures. The information is presented in Table 9.

This analysis allowed one to begin to make inferences. Hand movements were not found to be an interfering factor for most children. The atypical hand movements had not occurred yet when the gestures were expected to develop. Consequently the atypical hand movements probably did not interfere with the absent development of gestures.

Table 9 **ATYPICAL HAND MOVEMENTS AND GESTURE COMBINATIONS**

N=17
at.h.m.=Atypical hand movements

| Combinations: | Number of subjs. |
|---|---|
| No gestures & atypical hand movements (at.h.m.) before 13 months. | 5 |
| No gestures & at.h.m after 13 months. | 7 |
| Gestures before 13 months & at.h.m. before 13 months. | 0 |
| Gestures after 13 months & at.h.m. after 13 months. | 1 |
| Gestures before 13 months & at.h.m. after 13 months. | 4 |
| Gestures after 13 months & at.h.m. before 13 months. | 0 |

Another possible confounding variable interfering with the development of gestures is the delay in other areas of development. Parents were asked for information on when they noticed a delay or a reduction in gross motor, fine motor, eye-contact, and social interactions. The results are presented later in Table 11. The expected age range for the development of communicative gestures is 9 to 13 months. The mean age for noticing a delay in gross motor activities was 10.13 months and a fine motor delay of 12.72 months. Some delays were noticed as early as 5 months. It is possible that limited gross motor and fine motor abilities may have had an impact on the lack of

development of communicative gestures. Since only one question was asked for gross motor and fine motor abilities it is not possible to infer a relationship between the lack of gestures and declining gross and fine motor skills.

V. Parents' Comments: Part F

Parents were requested to comment on the following two questions:

1. Do you feel your child presently knows more than what she is able to express?
2. Is there anything in your child's communication development (understanding or expression) that you think might be important to share?

All comments are listed in Appendix M.

There were common themes that occurred in the parents' comments. Most parents stated that they felt their RS child knew more than she was able to express. Some explained this citing that their child gets excited and smiles when you ask her if she wants to do something she likes such as have a bath or go outside. Many felt that their child had definite preferences and dislikes. Others felt that their child used eye pointing or stood in front of something she wanted. Different types of screaming/ crying were felt to indicate excitement, pain, hurt feelings, fear, or frustration. Finally there were

comments about the children seeming to act as if they recognized happy versus sad music.

It appears that the parents' comments indicate that the children understand some contexts and situations and that they have some communicative intent. Once the regression has occurred the children may understand the overall context/situation but whether they understand the language or linguistic component is questionable.

Experimental Question 2: Extent of Regression.

Regression: (Part E)

Analysis of milestone regression is important to determine the extent of the regression and whether a stable point is reached where no further regression occurs. Regression (Appendix M) is described by comparing previously reported attained milestones (Part B questions) to the child's present level of milestones demonstrated (Part E). A total number of milestones lost were reported for each child in Table 10.

Table 10 **REGRESSION** N=17

| Age of subject: | Number of communication milestones attained: | Present number of milestones: | Number of milestones lost: |
|---|---|---|---|
| 23 mons. Y.D. | 5 | 5 | 0 |
| 27 mons. R.T. | 11 | 8 | 3 |
| 32 mons. L.E. | 11 | 10 | 1 |
| 33 mons. B.B. | 9 | 8 | 1 |
| 33 mons. O.N. | 7 | 9 | 0 |
| 35 mons. A.N. | 17 | 6 | 11 |
| 37 mons. R.Y. | 7 | 6 | 1 |
| 37 mons. N.O. | 7 | 7 | 0 |
| 41 mons. C.K. | 12 | 10 | 2 |
| 42 mons. N.N. | 13 | 8 | 5 |
| 46 mons. I.N. | 8 | 8 | 0 |
| 46 mons. N.S. | 7 | 3 | 4 |
| 47 mons. N.E. | 4 | 6 | 0 |
| 47 mons. A.M. | 16 | 5 | 11 |
| 47 mons. T.N. | 9 | 7 | 2 |
| 48 mons. G.E. | 15 | 8 | 7 |
| 48 mons. E.R. | 5 | 2 | 3 |

The results indicated that the majority of children in this study had already begun to regress. The extent of the regression in terms of lost milestones could not be determined unless these children were followed for a more extensive period of time. The older children in the study were presently at 2 to 8 milestones which corresponded to an approximate communication level of 2 to 8 months.

Experimental Question number 3: Other delays

Five questions focused on certain behaviors or noted delays in other areas of development. Reliability was sufficient to report the results for circling yes/no and then indicating an estimated age. The results were as follows in Table 11.

Table 11 **DEVELOPMENTAL DELAYS AND BEHAVIORS**

N=17

| | Yes: | No: | Mean: | S.D.: | Range: |
|---|---|---|---|---|---|
| A.First noticed your child appeared to "fall behind" in gross motor development (leg and arm movements such as crawling, walking, jumping etc.)? | 100% | 0% | 10.13 months | 4.91 months | 5-21 months |
| B. Noticed child appeared to "fall behind" in fine motor movements (ie. finger control)? | 100% | 0% | 12.72 months | 4.34 months | 6-20 months |
| C. Noticed child's attempts to seek human contact were decreasing (ie. reduction in frequency of approaching people)? | 82% | 18% | 14.33 months | 3.8 months | 8.5-20 months |
| D. Noticed child's solitary play with toys or objects was decreasing (ie. child's time spent playing with toys was less frequent)? | 76% | 24% | 16.13 months | 4.58 months | 8.5-24 months |
| E. Noticed child's eye-contact was less frequent than previously? | 71% | 29% | 16.23 months | 4.97 months | 6-24 months |

Some children presented with an earlier onset of developmental regression than others. The range for each

of the above questions was quite large with some noted delays as early as 5 months and some as late as 21 to 24 months. Delays in gross motor movements were noticed prior to other delays or behaviors.

# CHAPTER 5

## Discussion

This study should be viewed as a preliminary investigation into communication development and regression in children with Rett syndrome. A retrospective questionnaire was developed to explore early prelinguistic and linguistic communication development of these children. The results of this investigation revealed that early prelinguistic and linguistic communication development is delayed and altered in children who have RS. There is some variability in early communication development with a few children delayed from the onset with an early and abrupt arrest in further communication development. Most presented with scattered development to a highest attained level of anywhere from approximately 6 to 16 months. None of the children attained a communication level higher than approximately 16 months prior to regressing. The children who reached a milestone at the 13 to 16 month stage often presented with delays or unattained earlier milestones. In addition to delays or unattained communication milestones, most of the children did not use communicative gestures which are expected to occur between 9 and 13 months. The results suggest that the debilitating effects of the syndrome are evident in communication development as early as 6 weeks

in some children to up to as high as 16 months in other children.

This information may be useful for professionals to convey to parents to alleviate feelings of guilt associated with their child's delays and regression. If the syndrome is identified early professionals may be able to prepare parents that their child may lose some of their attained milestones. Most of the children in this investigation had lost milestones and were at a level where the following milestones were generally present: alerting, smiling, orienting, sometimes babbling. These milestones are at an approximate 2 to 8 month level. It could also be conveyed that at this point in time it is not possible to definitely determine what the child does and does not understand. On the comments section of the questionnaire, most parents felt that their RS child knew more than she was able to express in any volitional manner. Some comments were based on parents' interpretation of their child's emotional responses to situations and familiar yes/no questions or statements (crying, smiling/happy, frustrated). In addition, a few parents felt that their child was communicating what she wanted by moving herself to those places (eg. stood in kitchen, or in front of the T.V.). The comments indicated that some children may understand the situation/context and have intentional behavior, but they may not understand

the linguistic component or language itself. The parents' comments, however, may support the possibility that these children in addition to other difficulties are unable to motor-program purposeful behavior for speech, as well as fine and gross motor activities. Finding a way for researchers to investigate this is an enigma. Presently there does not seem to be a method or way to sort out to what extent motor programming is impaired and to what extent the difficulties are related to reduced cognitive abilities or comprehension.

Given the regression involved it may also be important for professionals such as speech-language pathologists to view maintenance of skills such as eye-contact and responsiveness to interactions (smiles etc.) as a priority over concentrating on attaining new skills/milestones.

If an etiology or medical treatment becomes known it would be crucial for earliest possible medical detection and intervention, as the syndrome's effects appear to result in delays at the onset or at a very early age in these children. It is also apparent that the regression in communication milestones and motor abilities begins prior to the most distinguishing feature of the syndrome, the atypical hand movements. Consequently often the regression is occurring before the syndrome is diagnosed. Presently there are no known early medical tests to detect the syndrome, leaving only reliance on observations of

regression and hand movements to detect the syndrome.

Though it is uncertain as to how the syndrome impairs the child's ability to communicate, it appears that communication development may not be "typical" or"normal" from the onset or from a very early age in children affected with this syndrome.

Limitations

The subjects for this investigation were the parents of the 17 Rett syndrome children. The conclusions are at best guarded due to the small number of subjects and the retrospective nature of the study.

A limitation to the investigation was that there were no known guidelines to determine if the minimum three week period prior to mailing a second questionnaire for the reliability analysis was an appropriate time frame. The estimated minimum four weeks between the two completions of the questionnaire may or may not have been a sufficient time for parents to forget how they responded on the questionnaire the first time. Reliability was not obtained on some measures which would suggest that the minimum four week period may have been sufficient between completions of the questionnaire, however, time frames could be investigated further. Parents were also forewarned that they would receive a second copy of the questionnaire and to please not retain a copy of the first questionnaire. This procedure was felt to be an ethical

necessity to guarantee subjects were adequately informed prior to authorizing consent.

The retrospective nature of the study was also a limitation. A prospective study would not have to rely entirely on parent recall and an independent observer could verify observations of attained milestones. Given that diagnosis does not occur at birth a prospective study is not feasible at this time.

## Research Implications

Based on the findings in this investigation there are several areas that could be explored in further research.

This investigation focused on the forty youngest children registered with the International Rett Syndrome Association. The subject sample was only 17 and consequently it would be useful to see if the results could be replicated with a larger N or with subjects from another country.

Given that parents commented that they felt that their child understands more than she is able to express and because of the possibility of apraxia limiting any type of physical or motor response, research in the development of techniques that might allow for the investigation of physiological responses to speech or situations would be useful. A device similar to a polygraph which measures a stress related response might be an area to explore. One

might be able to explore further just what these children understand if there are advancements in the development of appropriate instruments.

The section of the questionnaire that requested an age estimate as to when other areas of delay were noticed revealed that a delay in gross motor development was an area usually observed prior to other noted delays. Motor milestones in children with RS have been reported, but further exploration of gross and fine motor milestones may be beneficial.

Parents were requested to indicate which of six atypical hand movements occurred, and if observed, whether they occurred before of after 13 months. Most parents indicated these occurred after 13 months. Since hand movements are the single most important factor in differentiating Rett syndrome from other syndromes or conditions the hand movements could be explored in further detail. The six hand movement indicators from this investigation could be investigated to determine what their order or progression is and at what point in time these are noticed.

Language and development studies could focus on further norm referencing and validation of the milestones on the Clinical Linguistic Auditory Milestone Scale as well as developing valid norm referenced scales for measuring early communicative gestures.

Another area to explore would be whether there were differences in parents' ability to recall milestone information about their child among parents of normal, mildly delayed, moderately delayed, and severely to profoundly delayed children. Investigators could follow children prospectively and then at a later point have parents complete a retrospective questionnaire on the milestones. Would parents of moderately delayed children be more reliable at reporting early milestone information than parents of mildly delayed or normal children? Does reliability become worse with a longer recall period? For example, are parents with two year old children more reliable than parents of three year old children at recalling milestones for the birth to two year stage? Do other factors such as the child's birth order or number of children in the family affect reliability of parent recall of milestones? Many professionals request early milestone information on case history forms, yet there is no known information that would suggest that this is a reasonable task for parents to undertake or that they can provide reliable or accurate information.

Further inquiries into communication, motor, and hand movements in children with Rett Syndrome as well as further analysis of our methods and techniques that would allow us to reliably evaluate these areas are needed.

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## Appendix A

Rett Syndrome: Diagnostic Criteria

Necessary Criteria:

1. Apparently normal prenatal and perinatal period
2. Apparently normal psychomotor development through the first six months
3. Normal head circumference at birth
4. Five months to four years-Deceleration of head growth.
5. Six to 30 months-Loss of acquired purposeful hand skills, temporally associated with communication dysfunction and social withdrawal
6. Development of severely impaired expressive and receptive language, and presence of apparent severe psychomotor handicap
7. Stereotypic hand movements such as hand wringing/squeezing, clapping/tapping, mouthing, and washing-rubbing automatisms appearing after purposeful hand skills are lost
8. One to four years-Appearance of gait apraxia and truncal apraxia-ataxia
9. Diagnosis tentative until age two to five years of age

(Typical Rett syndrome diagnosis involves meeting the first eight criteria.)

Supportive Criteria

1. Breathing dysfunction
   a. Periodic apnea during wakefulness
   b. Intermittent hyperventilation
   c. Breath-holding spells
   d. Forced expulsion of air or saliva
2. EEG abnormalities
   a. Slow waking background and intermittent rhythmic slowing (3-5 Hz)
   b. Epileptiform discharges, with or without clinical seizures
3. Seizures
4. Spasticity often with associated development of muscle wasting and dystonia
5. Peripheral vasomotor disturbances
6. Scoliosis
7. Growth retardation
8. Hypotrophic small feet

Exclusion Criteria are mentioned as well.

Note. Modified slightly from "The Clinical Recognition and Differential Diagnosis of Rett Syndrome" by E. Trevathan and S Naidu, 1988, Journal of Child Neurology, 3(Suppl) p. S6-S16, Copyright 1988. Trevathan and Naidu (1988) listed modified information from Hagberg et al. (1985).

Appendix B

Rett Syndrome: Four Clinical Stages

| Stages | Clinical Characteristics |
|---|---|
| Stage I<br>Onset:<br>6-18 mos<br>Duration:<br>months | Deceleration of head/brain growth<br>Developmental stagnation<br>Disinterest in play/environment<br>Hypotonia EEG background: normal or minimal slowing of posterior rhythm. |
| Stage II<br>Onset:<br>1-3 years<br>Duration:<br>weeks to months | Rapid development regression/irritable<br>Insomnia. Seizures. Loss of hand use.<br>Hand stereotypes: wringing, clapping, mouthing. Self-abusive behavior (eg. chewing fingers, slapping face, pulling hair) Autistic manifestations<br>Loss of expressive language<br>EEG: background slowing, gradual loss of normal sleep activity; focal or multifocal spike/wave. |
| Stage III<br>Onset:<br>2-10 years<br>Duration:<br>mos. to years | Severe mental retardation/dementia<br>Amelioration of autistic features.<br>Seizures. Bruxism. Early Scoliosis.<br>Typical hand stereotypes: wringing, tapping, mouthing. ataxia and apraxia.<br>Hyperreflexia and progressive rigidity.<br>Hyperventilation, breath-holding, aerophagia during waking<br>Weight loss with excellent appetite.<br>EEG: gradual disappearance of posterior rhythm, generalized slowing, absent vertex and spindle activity,epileptic abnormalities activated during sleep |
| Stage IV<br>Onset:<br>10+years<br>Duration:<br>years | Scoliosis, muscle wasting, rigidity<br>Trophic disturbance of feet<br>Decreasing mobility, wheelchair-bound<br>Growth retardation. Improved eye contact. Virtual absence of expressive/receptive language<br>Reduced seizure frequency<br>EEG: poor background organization with marked slowing and multifocal spikes and slow spike and wave pattern in sleep |

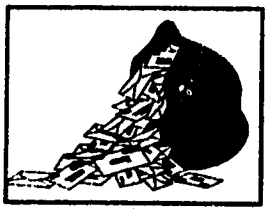
Note Modified slightly from "The Clinical Recognition and Differential Diagnosis of Rett Syndrome" by E. Trevathan and S. Naidu, 1988 Journal of Child Neurology, 3(suppl), p. S6-S16. Copyright 1988. Trevathan and Naidu (1988) listed modified from Hagberg and Witt-Engerstrom (1986).

# Appendix C

| Milestone: | Normative Population (N=381) Age (mons) Mean: | SD: | C var: |
|---|---|---|---|
| Alert (wks) | 1.1 | 1.3 | 1.21 |
| Smile (wks) | 5.0 | 2.2 | 0.44 |
| Coo (wks) | 6.5 | 2.7 | 0.41 |
| Orient (voice) | 2.8 | 1.2 | 0.32 |
| Ag-goo | 4.0 | 1.6 | 0.39 |
| Razz | 4.4 | 1.6 | 0.36 |
| Babble | 6.3 | 1.4 | 0.23 |
| Mama/dada indiscriminately | 7.7 | 1.7 | 0.22 |
| Gesture (ie. bye-bye) | 8.6 | 1.5 | 0.18 |
| Dada discriminantly | 10.5 | 2.5 | 0.24 |
| Mama discriminantly | 11.1 | 2.7 | 0.24 |
| One step command with gesture | 11.1 | 1.7 | 0.15 |
| First word | 11.3 | 2.3 | 0.20 |
| Immature jargon | 12.2 | 2.1 | 0.17 |
| Second word | 12.4 | 2.2 | 0.18 |
| Three words | 13.2 | 2.2 | 0.17 |
| One step command without gesture | 13.6 | 2.1 | 0.15 |
| Four to six words | 14.7 | 2.5 | 0.15 |
| Mature jargon | 16.5 | 2.9 | 0.18 |
| Five body parts | 16.7 | 2.8 | 0.17 |
| Seven to 20 words | 16.9 | 2.9 | 0.17 |
| Eight body parts | 19.0 | 3.2 | 0.17 |
| Two word combinations | 19.2 | 3.0 | 0.16 |
| Two word sentences | 20.6 | 3.0 | 0.15 |
| 50 words | 20.9 | 3.2 | 0.15 |

Note Modified from Table III from the "Clinical Linguistic and Auditory Milestone Scale: Prediction of Cognition in Infancy" by A. J. Capute, F. B. Palmer, B. K. Shapiro, R. C. Wachtel, S. Schmidt, & A. Ross, 1986, Developmental Medicine & Child Neurology, 28, 762-771. Copyright 1986.

Appendix D



CANADIAN QUESTIONNAIRE ON
EARLY COMMUNICATION DEVELOPMENT
IN CHILDREN WITH RETT SYNDROME

It is recognized that this questionnaire may take your time, patience, and recall of events that may be difficult for you to remember. Please feel free to use any resources available to you, such as a baby book, medical records, or talking with a family member.

I would like to thank you in advance for your time and efforts. It is hoped that the information will provide a clearer picture of early communication development in children with Rett syndrome.

Please circle or list the information for each question. Provide the answer closest to your best estimate.

**PART A**

Your name and address will remain strictly confidential. We are requesting that you provide your name and address as the research project will involve having a proportion of people complete the questionnaire a second time. If you are uncomfortable with this request you may leave these two questions blank.

Name of parent completing the questionnaire:

Address _____

_____

_____

City Province

_____

Postal Code

Child's date of birth: Year_____ Month_____Day_____

Biological mother's age_____ Biological father's age_____

Mother's highest education level attained_____
Mother's occupation_____

Father's highest education level attained_____
Father's occupation_____

Child lives in (circle): 1 2 3
Canada United States Other country

Child's age when diagnosed with Rett syndrome:
_____months old.

Child's sex: Circle 1 2
Female Male

Diagnosis confirmed by a physician (circle): Yes No

If no. List occupation of professional that provided diagnosis or confirmed diagnosis (for example a psychologist, etc.): _____.

If the diagnosis is questionable please explain:

_____

_____

_____

**PART B**

The next items require you to recall information for the first years of your child's life. Each item is an early communication behavior. Please respond to each item by placing a checkmark ( ) by either "observed" or "not observed". If the behavior was observed circle your best estimate of the age when you first observed it.

Please note that the first few questions refer to weeks and then the remaining questions refer to months.

1. Infant recognized the presence of sound by blinking, starting, and moving any part of the body?

Please check one: observed_____ not observed_____
If observed, please circle child's age when it was first observed (A,B, or C):

Child's Age: **A** **B** **C**
Before 2 1/2 weeks 2 1/2 to 3 1/2 weeks After 3 1/2 weeks

2. Infant smiled at you when you talked to her or stroked her face.

Please check one: observed_____ not observed_____
If observed, please circle child's age when it was first observed (A,B,or C):

Child's Age: **A** **B** **C**
Before 7 weeks 7 to 9 1/2 weeks After 9 1/2 weeks

3. Infant produced long vowel sounds in a musical fashion? Cooing.

Please check one: observed_____ not observed_____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 9 weeks | 9 to 12 weeks | After 12 weeks |

4. You entered a room and the baby didn't see you at first, she began to turn immediately to the correct side when you spoke to her rather than searching in the wrong direction at first?

Please check one: observed_____ not observed_____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 4 weeks | 4 to 5 weeks | After 5 weeks |

5. Baby first said "ah-goo"?

Please check one: observed_____ not observed_____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 5 1/2 weeks | 5 1/2 to 7 weeks | After 7weeks |

6. Baby first gave you a 'raspberry' sound (air blowing through lips with lips vibrating)?

Please check one: observed_____ not observed_____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 6 weeks | 6 to 7 1/2 weeks | After 7 1/2 weeks |

7. Infant first babbled (demonstrate repetitive strings of consonants eg."baba ba ba")?

Please check one: observed_____ not observed_____
If observed, please circle child's age when it was first observed:

Child' Age **(Note change to MONTHS)**:

| A | B | C |
|---|---|---|
| Before 7 1/2 **months** | 7 1/2 to 9 months | After 9 months. |

8. Infant first waved bye-bye or played pat-a-cake?

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

Child's Age:

| A | B | C |
|---|---|---|
| Before 10 months | 10 to 11 1/2 months | After 11 1/2 months. |

9. Child first said 'dada' and 'mama' but without reference to mother or father. Said 'dada' and 'mama' indiscriminately?

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

Child's Age:

| A | B | C |
|---|---|---|
| Before 9 1/2 mos. | 9 1/2 to 11 mos. | After 11 mos. |

10. Child first referred to the mother saying "mama"?

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

Child's Age:

| A | B | C |
|---|---|---|
| Before 13 1/2 mos. | 13 1/2 to 16 1/2 mos. | After 16 1/2 mos. |

11. Child first referred to the father saying "dada"?

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

Child' Age:

| A | B | C |
|---|---|---|
| Before 13 mos. | 13 to 15 1/2 mos. | After 15 1/2 mos. |

12. Child learned to say her first word other than "dada", "mama", or family names?

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 13 1/2 mos. | 13 1/2 to 16 mos. | After 16 mos. |

If possible name word_____

13. Child learned to say two words other than "dada", "mama", or family names? This means able to say a total of two words. It doesn't mean two words together in a sentence (eg. child said "hot", and later in day the child said "more").

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

| child's Age: | A | B | C |
|---|---|---|---|
| | Before 14 1/2 mos. | 14 1/2 to 16 1/2 mos. | After 16 1/2 mos. |

If observed, could you remember the first two words?___yes ___no
If possible list words 1._____
2._____

14. Child learned to say three words other than "mama", "dada", or family names? This means able to say a total of three words. It doesn't mean three words together in a sentence.

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 15 1/2 mos. | 15 1/2 to 17 1/2 mos. | After 17 1/2 mos. |

Name third word if possible 3._____

15. Child first followed simple commands such as "give me" or "bring me" accompanied by a gesture (ie. parent said "bring comb" while pointing to the bathroom.)?

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 12 1/2 mos. | 12 1/2 to 14 1/2 mos. | After 14 1/2 mos. |

16. Child first followed simple commands such as "give me" or "bring me" <u>not</u> accompanied by a gesture?

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 15 1/2 mos. | 15 1/2 to 17 1/2 mos. | After 17 1/2 mos. |

17. Child learned to say 4 to 6 words other than "mama", "dada", or family names. The words could have been said separately They did not have to be in a phrase or sentence.

Please check one: observed____ not observed____

If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 17 mos. | 17 to 19 1/2 mos. | After 19 1/2 mos. |

If possible name them :_____ _____
_____ _____
_____ _____

18. Child had a seven to twenty word vocabulary (able to say 7 to 20 words)? The words could have been said separately. They did not have to be in a sentence.

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 19 1/2 mos. | 19 1/2 to 22 1/2 mos. | After 22 1/2 mos. |

19. Child used jargon (unintelligible words) run together in a way that sounded like a sentence (eg. "abba deo dappi omma")?

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 14 mos. | 14 to 16 1/2 mos. | After 16 1/2 mos. |

20. Child's jargon began to include recognizable words (eg. "abba amma truck")?

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 19 1/2 mos. | 19 1/2 to 22 1/2 mos. | After 22 1/2 mos. |

21. Child was able to understand and point to five body parts?
Example: parent says "show me your nose" and child points to her nose.

Please check one: observed____ not observed____
If observed, please circle child's age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 19 1/2 mos. | 19 1/2 to 22 mos. | After 22 mos. |

22. Child was able to understand and point to eight body parts?

Please check one: observed____ not observed____
If observed, please circle age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 22 1/2 mos. | 22 1/2 to 25 1/2 mos. | After 25 1/2 mos. |

23. Child started to put two words together in a phrase (Not necessarily a sentence, frequently both nouns eg. "door truck")?

Please check one: observed____ not observed____
If observed, please circle age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 22 mos. | 22 to 25 mos. | After 25 mos. |

24. Child learned to say 50 words?

Please check one: observed____ not observed____
If observed, please circle age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 24 mos. | 24 to 27 1/2 mos. | After 27 1/2 mos. |

25. Child put a noun and verb together in a sentence (eg. "want juice" "push car")?

Please check one: observed____ not observed____
If observed, please circle age when it was first observed:

| Child's Age: | A | B | C |
|---|---|---|---|
| | Before 23 1/2 mos. | 23 1/2 to 26 1/2 mos. | After 26 1/2 mos. |

26. Child said "Wh" questions (eg. "What is that?", or "Where is that?")?

Please check one: observed____ not observed____
If observed, please circle age when it was first observed:
Child's Age: **(Note only two choices A or B)**

| A | B |
|---|---|
| Before 34 months. | After 34 months |

27. Child said "don't" or "can't" in sentences?
(eg. "He can't play.")

Please check one: observed____ not observed____
If observed, please circle age when it was first observed:

| Child's Age: | A | B |
|---|---|---|
| | Before 41 months | After 41 months |

28. Child said helping verbs such as "will", or "am" in sentences to ask a question (eg."Will he bring it home?")?

Please check one: observed____ not observed____
If observed, please circle age when it was first observed:

| Child's Age: | A | B |
|---|---|---|
| | Before 45 months | After 45 months |

29. Child asked "why" questions (eg. Why is it hot?)
Please check one: observed____ not observed____
If observed, please circle the age when it was first observed:

| Child's Age: | A | B |
|---|---|---|
| | Before 52 months | After 52 months |

**PART C** Communicative gestures are sometimes used to indicate a child's wants or needs before the child can communicate well by talking. Please continue with the following questions.

1. Pointing- child used index finger to point toward something, and looked at you to see if you were looking at it.

Please check one: observed____ not observed____
If observed, please circle the age when it was first observed:

| Child's Age: | A | B |
|---|---|---|
| | Before 13 months | After 13 months |

2. Showing- While playing with a toy or object the child reached out to show the object to the parent(s)?

Please check one: observed____ not observed____
If observed, please circle the age when this first occurred:

| Child's Age: | A | B |
|---|---|---|
| | Before 13 months | After 13 months |

3. Giving- Child purposely gave objects (toys/things) by placing them in the parent(s) hand or lap?
Please check one: observed____ not observed____
If observed, please circle the age when it was first observed:

| Child's Age: | A | B |
|---|---|---|
| | Before 13 months | After 13 months |

**PART D**

Certain atypical hand behaviors have been reported as a characteristic in children with Rett syndrome. Please respond to the following statements:

1. Noticed a decrease or reduction in purposeful use of the hands (ie. less grasping or holding onto toys, less ability to manipulate objects such as a spoon or toy.)

Please check one: observed____ not observed____
If observed, please circle the age when this first occurred:
Child's Age: A B
Before 13 months After 13 months

2. Noticed a point in time where purposeful use of the hands was totally absent(ie. unable to grasp or hold onto objects).

Please check one: observed____ not observed____
If observed, please circle the age when this first occurred:
Child's Age: A B
Before 13 months After 13 months

3. Noticed hand(s) repetitively pulling or tapping on other parts of the body (ears, chest, or mouth, etc.)

Please check one: observed____ not observed____
If observed, please circle the age when this first occurred:
Child's Age: A B
Before 13 months After 13 months

4. Noticed hand(s) repetitively squeezing into a fist and then releasing.

Please check one: observed____ not observed____
If observed, please circle the age when this first occurred:
Child's Age: A B
Before 13 months After 13 months

5. Noticed hand(s) waving at sides of the body or in the air.

Please check one: observed____ not observed____
If observed, please circle the age when this first occurred:
Child's Age: A B
Before 13 months After 13 months

6. Noticed hand wringing behavior.
Please check one: observed____ not observed____
If observed, please circle the age when this first occurred:

| Child's Age: | A | B |
|---|---|---|
| | Before 13 months | After 13 months |

It is of interest to determine whether the atypical hand behaviors (Part D) interfered with the development of the communicative gestures of giving, showing, pointing (Part C questions).

I. If you observed your child use a communicative gesture (Part C) and observed atypical hand movements (Part D), **both** before or both after 13 months, please give us some estimate of which type of behaviors occurred first. Circle:

| A | B | C |
|---|---|---|
| communicative gestures occurred **before** atypical hand movements | atypical hand behaviors occurred **before** communicative gestures | atypical hand movements and gestures occurred at about the same time |

We are trying to understand the relationship between language and other areas of development. Please read the following statements. Circle yes or no to indicate if it was observed or not. If circled yes please try to estimate an age.

A. First noticed your child appeared to "fall behind" in gross motor development (leg and arm movements such as crawling, walking, jumping etc.). yes no If yes age __

B. Noticed child appeared to "fall behind" in fine motor movements (ie. finger control). yes no If yes, age__

C. Noticed child's attempts to seek human contact were decreasing (ie. reduction in frequency of approaching people). yes no If yes, age__

D. Noticed child's solitary play with toys or objects was decreasing (ie. child's time spent playing alone with toys was less frequent). yes no If yes, age__

E. Noticed child's eye-contact was less frequent than previously. yes no If yes, age__

**Part E** Please place a check mark beside any of the following behaviours that your child is **PRESENTLY** doing. These are the same questions that were asked in part B; however we are now wanting to know what your child is presently doing. If some of the following are not clear to you please return to read them again in part B.

1. Alerts to sound ☐ 2. Smiles when talked to ☐

3. Coos ☐ 4.Turns to orient to voice ☐ 5.Says ah-goo ☐

6. Makes a raspberry sound (blows air through lips) ☐

7. Babbles (eg. ba ba ba) ☐ 8. Gestures ( eg. waves bye) ☐

9. Says mama dada but not referring to the correct person ☐

10.Says mama referring to mom ☐ 11. Says dada referring to dad ☐

(For questions 12 to 16 words other than "mama", "dada", or family names, and the words can be said separately, not necessarily in a sentence)

12. Says one word ☐ 13. Says two words ☐ 14. Says three words ☐

15. Says four to six words ☐

16. Says 7 to 20 words ☐ 17. Says 50 words ☐

18. Follows a direction if you also use a gesture (eg. "up" while holding out hands) ☐

19. Follows a direction without a gesture (eg. "up" but without holding out hands) ☐

20. Uses jargon -a run of unintelligible words (eg. akka alla bobob de) ☐

21. Uses jargon with some intelligible words (eg. bidda daba car) ☐

22. Knows and points to 5 body parts ☐

23. Knows and points to 8 body parts ☐

24. Uses two word phrases (eg. "ball floor") ☐

25. Uses two words, a noun and a verb (eg. "push me") ☐

26. Uses words "can't, don't won't" in sentences (eg. "He can't play.") ☐

27. Uses words "will", "am" to ask a question (eg. Will he bring it home?) ☐

28.Uses wh questions in sentences (eg. Where is it? What is it?) ☐

29.Uses "why questions"("Why is he running?") ☐

**Part F**

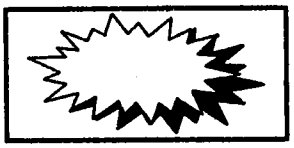
**Do you feel your child presently knows more than what she is able to express?**
**Circle: Yes or No**
**Please explain:**_____

_____
_____
_____
_____
_____
_____
_____
_____
_____
_____

**Is there anything in your child's communication development (understanding or expression) that you think might be important to share? Please feel free to comment:**

_____
_____
_____
_____
_____
_____
_____
_____
_____
_____

**PLEASE TURN BACK THROUGH THE QUESTIONNAIRE TO CHECK THAT ALL QUESTIONS HAVE BEEN ANSWERED.**



Thank you for your time.

## Appendix E

(Canada)
Dear subscriber of the Canadian Rett Syndrome Ass. Newsletter:

Hello, my name is Shelley Little and I am a certified speech-language pathologist working in the province of Alberta. Presently, I am also working on the completion of my master's degree in the department of Speech Pathology and Audiology at the University of Alberta. My thesis research project is on the Rett syndrome, and is a requirement for my master's degree.

My interest in Rett syndrome initially grew out of clinical experience in working with a child with Rett syndrome. I am using a questionnaire to document information on communication development for the early years of life of children with Rett syndrome.

Health professionals rarely see the children with Rett syndrome before the late infancy or the preschool years. As a result, critical information is lacking for the early years of development.

The results of this study will provide helpful information about these children's early communication development and may increase health professionals' awareness and understanding of Rett syndrome.

Please note this questionnaire was provided to the Canadian Rett Syndrome Association for them to mail to subscribers of the Newsletter. The principal investigators will not know your name until you complete and return the questionnaire. If you are not interested in completing the questionnaire there is a blue form enclosed for you to check. Please see the blue page, place a check mark where appropriate, and return the blank questionnaire. Returning the questionnaire is important so that we may determine an accurate return rate. If you choose to complete the questionnaire, please read and sign the consent form, and then complete the questionnaire. Keep a copy of the consent form, and return the enclosed questionnaire and consent form in the self addressed envelope. Please note for convenience the questionnaire will be returned to me at my place of employment at the Red Deer Regional Health Unit.

If you are aware of a family who does not subscribe to the Rett syndrome Association Newsletter feel free to inform them of the questionnaire. If they are interested they can contact me and I will send them a questionnaire.

Your consideration and efforts are greatly appreciated. Thank you in a advance for your time.

Yours truly,

Shelley Little, B.Sc. (c)
Registered Speech-Language Pathologist

Appendix F

(Canada)

CONSENT TO PARTICIPATE

Department of Speech Pathology and Audiology
University of Alberta.

EARLY COMMUNICATION DEVELOPMENT IN RETT SYNDROME

PRINCIPAL INVESTIGATOR: Shelley Little, Certified Speech-Language Pathologist
Work: 403-341-2160
THESIS SUPERVISOR: Dr. Gary Holdgrafer, Professor
University of Alberta
403-492-5980

The purpose of this research project is to study early communication development in children with Rett syndrome.

Information concerning the development of children with Rett syndrome is lacking, particularly for the early years of the child's life. The results of the questionnaire should provide valuable information to health professionals.

If you choose to take part in this research you are asked to complete the enclosed questionnaire regarding your child's early language development. It may be difficult to recall when your child achieved specific milestones such as babbling. Please feel free to use any resources available to you, such as, a baby book, medical records, or a videotape to help you respond to the questions.

The primary focus of this study is on children presently under the age of 10 years. If your child is older than ten years please see the blue check sheet, and then you may return the blank questionnaire so we are able to determine the response rate.

This research has been approved by the Ethics committee at the Department of Speech Pathology and Audiology at the University of Alberta. These packages containing the questionnaire have been mailed to you directly from the Rett Syndrome Association. Your identity will not be known to the principal investigators until you choose to complete and return the questionnaire. All identifying information will remain confidential ensuring your anonymity.

If you respond to the questionnaire, a proportion of respondents will be asked to complete the questionnaire a second time in order to determine how difficult it is for parents to remember information about their child's language development. You may be asked to complete a second copy of the questionnaire within eight weeks of

returning the initial questionnaire. The second copy may be useful to other investigators doing similar research to have a better understanding of how difficult this task is for parents.

You may decide not to participate for the questionnaires and you may refuse to answer any individual questions.

A general summary of this study will be offered to the Rett Syndrome Association Newsletter. If you have any questions, please feel free to contact Shelley Little at 403-341-2160. Dr. Holdgrafer can be reached at 403-492-5980.

By signing below, you acknowledge that you have been adequately informed about the purpose and possible future use of the research results. You understand that confidentiality will be maintained, and your rights not to participate. Your signature confirms your voluntary consent to participate and acknowledges your receipt of a copy of this informed consent letter. Please keep one copy of this consent form.

**Please return the signed consent form with the questionnaire.**

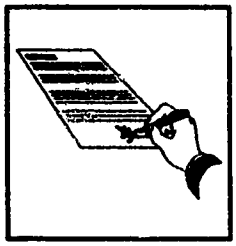
_____ _____

Parent/Guardian signature Date

_____ _____

Principal Investigator Date

Shelley Little, B.Sc. (c)
Registered Speech-Language Pathologist



## Appendix G

Dear member of the International Rett Syndrome Association:

Hello, my name is Shelley Little and I am a certified speech-language pathologist working in the province of Alberta in Canada. Presently, I am also working on the completion of my master's degree in the department of Speech Pathology and Audiology at the University of Alberta. My thesis research project is on the Rett syndrome, and is a requirement for my master's degree.

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Health professionals rarely see the children with Rett syndrome before the late infancy or the preschool years. As a result, critical information is lacking for the early years of development.

The results of this study will provide helpful information about these children's early communication development and may increase health professionals' awareness and understanding of Rett syndrome.

Please note this questionnaire was provided to the International Rett Syndrome Association for them to mail to some of their members. The principal investigators will not know your name until you complete and return the questionnaire. If you are not interested in completing the questionnaire there is a blue form enclosed for you to check. Please see the blue page, place a check mark where appropriate, and return the blank questionnaire. Returning the questionnaire is important so that we may determine an accurate return rate. If you choose to complete the questionnaire, please read and sign the consent form, and then complete the questionnaire. Please try to complete the questionnaire within 10 days. Keep a copy of the consent form, and return the enclosed questionnaire and consent form in the self addressed envelope. Please note for convenience the questionnaire will be returned to me at my place of employment at the Red Deer Regional Health Unit.

Your consideration and efforts are greatly appreciated. Thank you in a advance for your time.

Yours truly,

Shelley Little, B.Sc. (c)
Registered Speech-Language Pathologist

## Appendix H

### CONSENT TO PARTICIPATE

Department of Speech Pathology and Audiology
University of Alberta.

### EARLY COMMUNICATION DEVELOPMENT IN RETT SYNDROME

PRINCIPAL INVESTIGATOR: Shelley Little, Certified Speech-Language Pathologist & Graduate Student
Work: 403-341-2160
THESIS SUPERVISOR: Dr. Gary Holdgrafer, Professor
University of Alberta
403-492-5980

The purpose of this research project is to study early communication development in children with Rett syndrome.

Information concerning the development of children with Rett syndrome is lacking, particularly for the early years of the child's life. The results of the questionnaire should provide valuable information to health professionals.

If you choose to take part in this research you are asked to complete the enclosed questionnaire regarding your child's early language development. It may be difficult to recall when your child achieved specific milestones such as babbling. Please feel free to use any resources available to you, such as, a baby book, medical records, or a videotape to help you respond to the questions.

This research has been approved by the Ethics committee in the Department of Speech Pathology and Audiology at the University of Alberta. These packages containing the questionnaire have been mailed to you directly by the International Rett Syndrome Association. Your identity will not be known to the principal investigators until you choose to complete and return the questionnaire. All identifying information will remain confidential ensuring your anonymity.

If you respond to the questionnaire you will be asked to complete the questionnaire a second time in order to determine how difficult it is for parents to remember past information about their child's language development. You may be asked to complete a second copy of the questionnaire within six weeks of returning the initial questionnaire. The results from the second copy may be useful to other investigators doing similar research to have a better understanding of how difficult this task is for parents.

You may decide not to participate for the questionnaires and you may refuse to answer any individual questions.

A general summary of this study will be offered to the International Rett Syndrome Association. If you have any questions, please feel free to contact Shelley Little in Alberta, Canada at 403-341-2160. Dr. Holdgrafer can be reached at 403-492-5980.

By signing below, you acknowledge that you have been adequately informed about the purpose and possible future use of the research results. You understand that confidentiality will be maintained, and your rights not to participate. Your signature confirms your voluntary consent to participate and acknowledges your receipt of a copy of this informed consent letter. Please keep one copy of this consent form.

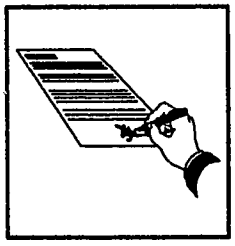
**Please return the signed consent form with the questionnaire.**

_____ _____

Parent/Guardian signature Date

_____ _____

Principal Investigator Date
Shelley Little, B.Sc. (c)
Registered Speech-Language Pathologist



# Appendix I

(Canada)

**Whether or not you decide to participate,** please check and return this form and the questionnaire.

**Please check one of the following:**

I received this questionnaire and I am the parent/guardian of a child with Rett syndrome. I am returning the completed questionnaire and consent form. ☐

I received this questionnaire but am <u>not</u> a parent/guardian of a child with Rett syndrome. I am returning the <u>blank</u> questionnaire so the questionnaire response rate can be determined. ☐

I received this questionnaire and I am the parent/guardian of a child with Rett syndrome but choose not to complete the questionnaire. My child is age _____. ☐

I received this questionnaire and I am the parent/guardian of a child with Rett syndrome. I am returning the blank questionnaire as my child is older than age ten. I am returning it so that you may determine the response rate. ☐

# Appendix J

(United States)

Whether or not you decide to participate, please check and return this form **and** the completed or blank questionnaire.

**Please check one of the following:**

I received this questionnaire and I am the parent/guardian of a child with Rett syndrome. I am returning the completed questionnaire and consent form. ☐

I received this questionnaire but am not a parent/guardian of a child with Rett syndrome. I am returning the blank questionnaire so the questionnaire response rate can be determined. ☐

I received this questionnaire and I am the parent/guardian of a child with Rett syndrome but choose not to complete the questionnaire. My child is age _____. ☐

Appendix K

**IMPORTANT SECOND LETTER. PLEASE READ CAREFULLY**

Date:

Name of subscriber and address

Dear _____:

Thank you for completing and returning the questionnaire. The data analysis for the questionnaires will begin once the second phase of the study is complete. As we told you in the initial consent form it is requested that parents complete the questionnaire a second time to determine how difficult it is for parents to remember information about their child's language development. This information itself will be useful to other researchers doing similar research to have a better understanding of how difficult recalling past information is for parents. I realize this would require at least a half hour of your time and I hope this is not too much of an inconvenience for you.

Please complete the enclosed questionnaire and return it. If you made a copy of the first questionnaire <u>please do not refer to it</u>. Do feel free to again use any information (baby book etc.) that will assist you in completing the questionnaire.

If you have any questions or concerns please feel free to contact me at 403-341-2181. Thank you again for your time.

Sincerely,

Shelley Little B. Sc. (c)
Registered Speech-Language Pathologist

## Appendix L
## Biographical Information

1. Child's Present Age:

| Subjects: | Age (months): |
|---|---|
| N.O. | 37 |
| L.E. | 32 |
| B.B. | 33 |
| N.N. | 42 |
| R.T. | 27 |
| C.K. | 41 |
| T.N. | 47 |
| I.N. | 46 |
| Y.D. | 23 |
| E.R. | 48 |
| O.N. | 33 |
| R.Y. | 37 |
| N.S. | 46 |
| A.M. | 47 |
| G.E. | 48 |
| N.E. | 47 |
| A.N. | 35 |

Range: 23-48 months
Mean: 39 months
S.D.: 7.9 months
Variance: 63

2. Child's age when diagnosed with Rett Syndrome.

| Subjects: | Age (months): |
|---|---|
| N.O. | 27 |
| L.E. | 24 |
| B.B. | 19 |
| N.N. | 24 |
| R.T. | 21 |
| C.K. | 30 |
| T.N. | 34 |
| I.N. | 27 |
| Y.D. | 23 |
| E.R. | 40 |
| O.N. | 26 |
| R.Y. | 32 |
| N.S. | 36 |
| A.M. | 34 |
| G.E. | 33 |
| N.E. | 29 |
| A.N. | 38 |

Range: 19-40
Mean: 29.2
S.D.: 6.1

3. Biological mother's age and Biological fathers age at time of birth.

Parents were requested to place their age and then the child's age was subtracted by the principal invetigator. It was felt that this question may have been misinterpreted by parents with some of them writing their age as their age when their child was born. Nevertheless the results are as follows:

| Subjects: | Biological mother's age at time of child's birth: | Biological father's age at time of child's birth: |
|---|---|---|
| N.O. | 36 | 36 |
| L.E. | 20 | 24 |
| B.B. | 23 | 28 |
| N.N. | 38 | 31 |
| R.T. | 20 | 20 |
| C.K. | 24 | 26 |
| T.N. | 30 | 31 |
| I.N. | 29 | 31 |
| Y.D. | 21 | 21 |
| E.R. | 19 | 19 |
| O.N. | 34 | 23 |
| R.Y. | 27 | 29 |
| N.S. | 31 | 33 |
| A.M. | 26 | 28 |
| G.E. | 22 | 29 |
| N.E. | 26 | 32 |
| A.N. | 31 | 35 |
| Range: | 19-38 | 19-36 |
| Mean: | 26.9 | 28.0 |
| S.D.: | 5.8 | 5.1 |

4. Mother's highest education level attained and mother's occupation:

The highest education level attained by mothers were as follows: 4 mothers high school, 11 college, and 2 university.

The occupations were as follows:

- 4 homemakers
- 3 students
- 1 sales manager
- 1 dental hygienist
- 1 dental lab technician
- 1 public relations
- 1 accountant
- 1 account coordinator
- 1 secretary
- 1 nurse's aide
- 1 nurse
- 1 sales

4. Father's highest education level attained and father's occupation:

The highest education level attained by fathers were as follows:
7 high school, 1 technical college, 5 college, 4 university.

The occupations were as follows:

- 1 highway labourer
- 1 student
- 1 millworker
- 1 contractor
- 1 unemployed
- 3 sales
- 2 managers
- 1 brake operator
- 1 programmer
- 1 bank collector
- 1 duty sheriff
- 1 aircraft mechanic
- 1 foreman
- 1 farmer

## Appendix M
## Section F

Section F:
Do you feel your child presently knows more than what she is able to express?

Parents' comments:

NN: [Child's age 42 months] Yes. She appears to be very socially aware; eg. If people in a room are laughing, she will too. She responds to her name by looking up and smiling. When using adapted toys, she responds to positive reinforcement and seems proud of herself. She knows positioning commands such as"sit" and "stand up"

RT: [Child's age 27 months] Yes. She gets a look in her eyes like she wants to communicate but is not able. She points to things with her eyes and gets very frustrated if she doesn't get what she wants. By her facial expressions and body gestures, I believe she knows and understands a lot more than she can communicate.

AM: [Child's age 47 months] Yes. She has a bright expression and is very alert most days. She uses her eyes to communicate in many ways. She seems to take in much more than her body allows her to give out. She will look into my eyes intently and it is obvious that she loves me. She recognizes people such as her grandparents whom she sees only once a month. She perks up when the bus arrives in the morning to take her to school. She has definite likes and dislikes and finds ways to tell us so. She remembers familiar places. Certain people make her burst into laughter whenever they appear. She looks so alert and seems to understand some phrases such as "lets go ride in the car" because her eyes light up.

CK: [Child's age 41 months] Yes. Sometimes She will do something out of the blue that surprises me. She makes it look as if she knew what she was doing. This happens once in a great while. I believe she knows what is going on around her and she recognizes people. You can see it in her eyes (so to speak) when she looks at objects she wants- she is eye pointing and it is consistent and obvious. When she looks at someone for a long time it is someone she has seen before or a voice she has heard before. She usually does not pay any attention to strangers when they talk to her buy smiles/laughs when she knows someone.

ER: [Child's age 48 months] Yes. I feel that she knows when you put a new outfit on her because she is usually happy. The way she looks at you sometimes and smiles when you talk to her.

ON: [Child's age 33 months] Yes. She becomes very animated in her body and facial movements under different circumstances. For instance when its time for going walking in the evening she becomes excited, kicking her legs waving her arms and perhaps fussing- as a baby would. I know when I ask her do you want a drink or take a bath- simple things- she understands-again through animated body movements. Her response to "drink", "bath", "outside" leads me to believe she knows- she becomes very animated. Just recently I've begun to see her shake her head back and forth in a "no" response when agitated. Also just 2 weeks or so ago she spoke the word "cow" (while in her wheelchair with 3 other adults present). We were out watering cattle at the time. She has also said "no". But has not said anything except babbling since those isolated times.

NO: [Child's age 37 months] Yes. I feel she understands more than she is able to express because if I ask her certain things she gets excited or her eyes light up and she smiles. Like do you want to take a bath, go bye-bye, want a drink, go outside? Sometimes she even shakes her head yes (up and down).

GE: [Child's age 48 months] Yes. She knows when someone is talking about her. She gets embarrassed and hides when people talk about her to me. She understands simple commands real well. If we ask her if she is thirsty or hungry she goes to the kitchen if she is thirsty or hungry. She is also very sensitive to others' moods or feelings.

LE: [Child's age 32 months] Yes. She has alot of nonverbal communication. She does alot of eye pointing. She can clearly make choices by eye pointing. She is in speech therapy and picks the same book every time when offered the books that she favours. She gets angry when she feels it is her time to be communicating with someone and she is not being communicated with. eg. When I pick her up from daycare and I talk to daycare providers after a while she slaps her hands on my chest and makes a vocal sound. She also does this at the beginning of speech therapy when the therapist and I are talking.

There are so many things times that it seems that she knows. It is a very difficult situation because she can't talk, walk, or use her hands appropriately so it is very hard to learn her system of communication.

BB: [Child's age 33 months] Yes. She seem to understand what is being said to her sometimes (I also feel that a lot is my imagination). She's always pleasant- there's never any harsh words spoken to her though. She loves to shop with me- loves it outside- she understands that she's somewhere other than home (I think).

TN: [Child's age 47 months] If you ask her if she wants a drink she will turn around and come back while you get it. She cries when her brother is in trouble (me or dad yelling at him) "Want to go bye-bye?" She gets excited also for "want to take a bath?" Very excited and heads to the bathroom. she loves her bath.

IN: [Child's age 46 months] Yes. Absolutely. She knows what's going on around her. She responds to questions and also her bath being run. She knows when you are talking about her and will smile at you.

YD: [Child's age 23 months] Yes.In feeding and if I ask her if she wants me to pick her up often she will turn away but is happy when I pick her up. Feeding, it seems that she wants to eat but can not make herself swallow when she wants to. She used to eat better.

RY: [Child's age 37 months] Yes. Communicated with eye pointing at times uses voice affliction in grunts and screams differently to communicate the following: excitement, pain, hurt feelings, fear, and frustration. In an unfamiliar environment she shuts down.
To "love" on something or to show interest she leans forward to touch it with her head to it.

NS: [Child's age 46 months] Yes. Eye contact; very observant.

NE: [Child's age 47 months] I don't know. Sometimes yes and other times no.

AN: [Child's age 23 months] Yes.

Part F Second Question:
Is there anything in your child's communication development (understanding or expression) that you think might be important to share?

NO: By the age of 20 months before she was diagnosed with Rett syndrome she was found to have a bilateral severe sloping sensorineural hearing loss. I sometimes wonder if it is a hearing loss or just a part of Rett syndrome where they sometimes act as if they are deaf. Even when she doesn't have her hearing aids in she still understands what we are saying (eg. want to take a bath, have a drink).

ON: Yes. Over a period of time she may show marked progress- perhaps in repetitive sounds for example- dada then she will stop after a few days and not do it again for weeks even months. She talks most when alone in her bedroom. She loves music and I've found that most helpful in times when she is upset or fretful.

ER: About the only way she communicates to us that she wants something is by crying.

CK: She has her own little TV with VCR in her room. When the video is finished when will start yelling (babble) until someone goes into her room to change or rewind the video then she will stop. This is also consistent. If she is in another room of the house and being real quiet- I will yell (from another area) "(NAME) are you O.K.?, and she'll babble something every time back her way of telling me she is O.K. If I ask her if she wants to take a bath or go swimming, she stares at the tub or pool. She definitely eye-points and that is our way of communication.

AM: She cannot get to standing from sitting, but when she's on her feet she can walk into the kitchen to "tell" us that she's hungry or she will stand in front of her video tape player to alert us to turn it on to her favourite tape. She fusses and whines if we put on the wrong video and giggles when we finally get it right. She clamps her mouth shut when we try to feed her the foods she dislikes. She will knock a box of crackers off the table. She can make the medicine she dislikes dribble out of her mouth. Sometimes when we ask for "love" and give her ample time she will lean over as if to hug us and even give us a wet kiss.

RT: Her eyes are her source of communication. If you are doing something or feeding her and quit, she'll hit you if she wants more.

NN: When she was about 11-12 months old, she could "ask" me to pick her up from her crib by saying "beka" for "pick up". She seems aware of where she is and communicates in tones. For example, she "complains" with a berating tone if her music has stopped.
She has definite preferences and will make them known by her sounds/tones.

RY: Strangely enough at times my child will blurt out 1 and 2 word phrases without warning in a store, in her room, different places but never repeats this word or 2 word phrase more than twice- not associated with medication, fever, or even having been coached to say these phrases ( eg. "thank you" "choo choo" "likes that" "birdie").
Using her head and mouth she can use a switch toy purposefully and laugh at the toys reaction. Therefore cause and effect has been demonstrated. Her eye contact in a quite room while being read a story is 100% better than in a crowded room.

YD: She never really developed any communication except by crying when she didn't like something. She had seizures (infantile spasms at a young age (approx. 4-5 months) and I feel this delayed her development at a very early age.

LE: She can also tell me if she is hungry. I ask her if she is hungry and she responds by making a squeal if she is hungry and nothing if she isn't. This is also true for going potty. She uses eye pointing on a regular basis. We use this form of communication for many things. She uses this form of communication to choose if she wants a bite of food or a drink at meal time. She use to pick a toy or book (two books choose one, two toys choose one). Sometimes she will pick what food she actually wants to eat. She picks clothes. According to the daycare provider and speech pathologist, she recently said the words up, stop, and kiss. In my opinion she knows more than she is able to express clearly. Just by the way she interacts and communicates nonverbally. She because of her extreme hand mouthing and finger twisting can not point or appropriately use her hands to communicate.
I find it hard to pin down to say when she does A it means B. It isn't as clear cut as that so it makes answering some of these questions difficult. Plus the fact that she once did also of these things it is difficult to remember when she did specific things.
BB: No.

TN: No.

IN: She makes her wants and needs known even though she has very few words. She can turn off the T.V. when she's not getting enough attention. She will come to us when she wants to be held or changed. Her communication is more than mere words.

NS: -

NE: She is more social than any Rett's child I've ever seen. Her social skills are improving with age.

AN: She is an extremely sensitive child, especially to music. A fast, happy, familiar tune will bring sounds of delight and big smiles. A slow, sombre, familiar tune (such as Silent Night) will bring tears in just a few words. She appears very bright and intense toward people like she understands everything but when it comes to expression that is where she fails.

She is very sensitive to happy or sad music.

GE: -