The Rett Syndrome Complex: Communicative Functions in Relation to Developmental Level and Autistic Features

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The Rett syndrome complex

Communicative functions in relation to developmental level and autistic features

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ABSTRACT Communicative functions and their relationship with overall developmental level and autistic features were studied in eight young women with disorders in the Rett syndrome complex, three with the classical variant and five with variants with partially preserved/regained speech. The Vineland Adaptive Behaviour Scales, individual structured observation, and a specially designed structured interview covering early pre-linguistic and linguistic development, current receptive and expressive language and non-verbal communication skills, were used. Autistic features were evaluated in accordance with the DSM-IV. Low levels of communicative abilities and overall functioning were demonstrated. In most cases, the communicative abilities did not reach the level expected on the basis of overall development. Joint attention behaviours and expressions of communicative intent were rare. However, six of the eight subjects showed clear examples of social interaction abilities. 'Eye pointing', as distinct from 'eye communication', was demonstrated only in a minority of the cases. The level of communicative function at developmental arrest did not predict later language ability. It is suggested that intervention should focus on developing further the joint attention behaviours, intentional communications and communicative functions spontaneously used by individuals with disorders in the Rett syndrome complex.

KEYWORDS *autism; communication; developmental level; language; Rett syndrome; speech*

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Rett syndrome (RS) represents a biologically odd and complex neurodevelopmental disorder (Percy, 1997), featuring severe loss of function between late infancy and the fourth year of life. Recent studies suggest a

gene locus at Xq28 (Amir et al., 1999). Male cases are extremely rare (Schanen et al., 1998). A diagnosis of RS is based on clinical consensus criteria (Hagberg et al., 1985; Trevathan et al., 1988). The course of the disorders may be divided into four characteristic stages: (1) early developmental stagnation; (2) severe psychomotor disintegration and regression; (3) partial cognitive and communicative recovery, but prominent deficit in motor planning; and (4) late gross motor deterioration (Hagberg and Witt-Engerström, 1986; Kerr 2000).

During the last decade, several atypical cases or variants of RS have been described and analysed by Hagberg and Skjeldal (1994). Atypical RS contrasts with classical RS in terms of either age of onset, or severity of symptoms, or both. The majority of atypical cases in this 'RS complex' (Zappella et al., 1998) present with milder symptoms and cannot be diagnosed with certainty before school age. Hagberg (1997) has suggested that the age of 10 years should be used as the lowest age limit for a conclusive diagnosis of RS variants. The best-known variants in the RS complex are forme fruste RS (comprising about 80 percent of atypical cases), congenital onset RS, infantile seizure onset RS, and the late regression variant. There is also a preserved/regained speech variant of RS. All the variants in the RS complex have been characterized by Hagberg and Gillberg (1993).

Neurological symptoms (including extreme dyspraxia) and severe linguistic and communicative difficulties are always present, and are included among the necessary criteria for a clinical diagnosis (Hagberg and Witt-Engerström, 1986). Autistic symptoms are common, but not required for diagnosis (Witt-Engerström and Gillberg, 1987). The characteristic initial development of girls with RS through infancy and very early childhood parallels that of children in general. Therefore, critical developmental transitions in pre-linguistic development will be highlighted. Such transitions are also relevant for the understanding of the emergence of autistic features.

The developmental course of linguistic and communicative abilities in children without major disabilities has its roots in infant pre-verbal communication; even new-born babies demonstrate social intent and active interest in communicating (Bateson, 1975; Murray and Trevarthen, 1986). The next step in development encompasses communication for information purposes. The child uses communicative acts like pointing or reaching out to show or to request objects. So-called 'joint attention', the act where two or more persons coordinate their attention to a common focus of interest, is an important part of this informative communicative function. Making eye contact and following the gaze of others to discover the other's focus of interest ('eye-direction detection') are behaviours necessary for the development of joint attention. Joint attention behaviours also

play a central role in identifying gestures as intentionally communicative. Early in development, caregivers strengthen the child's first attempts at intentional communication by attributing intent to all kinds of communicative behaviour, even if there is no such awareness on the child's part. Intentional communicative behaviour implies taking account of the communicating partner, attracting the other's attention and directing it to the communicative target, and also understanding that no communicative signal can work if the addressee does not perceive it. Intentional communication and the special case of use of repair strategies to clarify messages clearly indicate an emerging 'theory of mind' or an ability to understand that persons to whom you send messages perceive the same messages, understand them (or not) and subsequently act upon them (Mundy and Willoughby, 1998; Wetherby et al., 1998). Menyuk (1975) showed that infants already at 4 months of age possess parts of this competence, which includes the ability to take the other person's perspective in order to establish a common ground (Markova, 1995). By 4 years of age the child exhibits a more adult-like ability to take another person's perspective or a developed 'theory of mind' (Wimmer and Perner, 1983). Most of these behaviours are rarely seen in children with autism and the absences of some of them are among DSM-IV defining features of autistic disorder.

One of the first studies of verbal language in RS girls was by Hagberg and Rasmussen (1986), who reported a case of forme fruste RS regaining some speech after the regression period. Later studies of linguistic and communicative abilities in girls with RS show two main types of development. On the one hand, Skjeldal et al. (1995) described a girl who used long sentences before onset of symptoms, when her verbal speech was completely lost. From the use of several words at 8 months, her speech was reduced to babbling at 2 years of age. At $3\frac{1}{2}$ years she had regained some spoken language and at 5 she expressed needs and some preferences, even in short sentences. Burford and Trevarthen (1997) argued that communication in girls with RS becomes increasingly disorganized during the first year, while the girls retain social responsiveness, indicated by orientation towards faces and contact smiling, though without any sign of joint attention. In the studies by Woodyatt and Ozanne (1992a; 1992b; 1993; 1994; 1997) linguistic functions of girls with RS were lost at age 1 to 4 years. Some of the girls had used up to three words before onset. In most cases verbal speech was not regained. Also Sörensen and Viken (1995) reported normal development during the first year in a girl with RS. From 9 months of age, she used single words that developed into short sentences. At 18 months, she lost her abilities, but later regained them at approximately 4 years. She produced sentences of two to three words and responded verbally to communication by others. In contrast, Tams-Little and Holdgrafer (1996) found

communicative development to be deviant already before developmental arrest.

On the other hand, a late onset of speech without subsequent regression occurred in a number of cases reported by Zappella (1992; 1994). The girls said their first words at between 15 months and 6 years. At the ages of 11, 20 and 20 years respectively they used fully formed sentences of up to 10 words, sometimes echolalic and with pronoun reversals. Recently, Zappella (1997) reported that five out of eight cases showed the more characteristic course of early loss of language and regained speech ability during the recovery phase.

As far as receptive and expressive abilities were concerned, Zappella (1997) reported somewhat better verbal comprehension than expressive functions, while Woodyatt and Ozanne (1992a; 1992b; 1993) and von Tetzchner (1997), studying 42 cases, found receptive and expressive abilities to be at equally low levels. Some of the girls used a few words, but von Tetzchner (1997) argued that the use was more 'expressive' than truly symbolic, i.e. without reference to the named object or event. At 18 months, one of four girls with RS in the study by Sigafoos et al. (1996) lost three spoken words, which she had used for some time. The girls demonstrated some comprehension, e.g. reacted to their own names, but virtually no expressive speech apart from a few vocalizations.

Some of the girls used both rudimentary oral, vocal and gestural expressions (Burford and Trevarthen, 1997) and non-verbal means like signing, gestures, noises, touch and eye contact (Woodyatt and Ozanne, 1992a; 1992b; 1993). Most girls showed contact behaviours (Burford and Trevarthen, 1997) but communicative intentions and joint attention of objects of common interest were virtually absent (e.g. von Tetzchner, 1997; Woodyatt and Ozanne, 1992a; 1992b; 1993). Although the most common picture was that caregivers assigned meaning to the girls' goal-directed behaviour (Woodyatt and Ozanne, 1992a; 1992b), Sörensen and Viken (1995) reported that their subject produced some sounds with communicative intent.

Woodyatt and Ozanne (1994) highlighted the impact of oral and verbal dyspraxia on language development in four girls with RS. Previously, Witt-Engerström (1990), Lindberg (1991) and Budden (1993) had suggested oral dyspraxia (i.e. deficits in motor planning) as a factor interfering with speech in RS. Fontanesi and Haas (1998) also hypothesized that cognitive ability might be misjudged in RS because of dyspraxic difficulties and extremely prolonged responses to cognitive tasks. The findings of Woodyatt and Ozanne, however, suggested cognitive level to be the major factor explaining the speech and communicative ability in RS. In particular, the lack of motivation and severe attention deficits seemed to interfere with

learning ability in RS. However, the findings also suggested that cognitive abilities in girls with RS, who have developed sufficient cognitive skills to be able to demonstrate intentional communication, might be underestimated owing to physical inability to coordinate hand movements and oromotor movements for speech (Woodyatt and Ozanne, 1994).

There is little systematic knowledge about cognitive functioning in RS. Previous research indicates that girls with classical RS function at a level indicating severe to profound mental retardation (Fontanesi and Haas, 1998; Garber and Veydt, 1990; Olsson and Rett, 1987; van Acker, 1991; von Tetzchner et al., 1996). Problems of finding assessment methods adapted to the girls' specific difficulties must, however, be taken into consideration when interpreting these results.

Autistic behaviours, i.e. the symptom triad comprising restriction in the ability to engage in reciprocal social interaction, restriction in verbal and non-verbal reciprocal communication, and restricted imagination with repetitive and stereotyped behaviours, are common in RS. The social withdrawal and aloofness presenting along with a general disintegration of skills in the regression stage of RS may be acute and alarming. However, loss of social function may be a transient phenomenon, and after the regression phase, in many cases social interaction ability slowly improves over the years (Hagberg and Witt-Engerström, 1986; Kerr, 2000), even though there may be little evidence of communicative intent (Burford and Trevarthen, 1997; von Tetzchner, 1997; Woodyatt and Ozanne, 1992a; 1992b; 1993). In some cases, however, severe autistic behaviour persists after the age of 20 years (Zappella et al., 1998). It has to be spelled out that autism also shows positive development over time with regard to social interaction. While most children with autism are mute by age 3 years, many talk at age 6 years, while others will remain silent for the rest of their lives.

In summary, most studies have found the communicative and linguistic abilities of girls with disorders in the RS complex to be at a pre-verbal and pre-intentional level. Some of the studies have indicated that the communicative abilities are consistent with overall cognitive level (von Tetzchner et al., 1996; Woodyatt and Ozanne, 1992a; 1993; 1994).

The aim of the present study was to investigate the interrelationships between communication, cognition and autistic features in young women with one of two variants of RS complex disorders: classical RS and forme fruste RS. Specifically, we wanted to evaluate (1) linguistic and communicative development at the time of stagnation, (2) developmental course, including possible loss and reappearance of verbal speech abilities, (3) current use of communicative functions, especially joint attention and intentional communication, in relation to cognitive level, (4) presence of autistic features, and (5) the possible interactions between these variables.

Method

Participants

Eight girls and young women with classical RS or forme fruste RS, aged 11 to 36 years (mean 23 years), were recruited for the study. All participants were originally diagnosed and clinically followed up for many years by the third author (BH). Baseline clinical data are presented in Table 1. All participants were also rediagnosed according to RS variant criteria (Hagberg and Skjeldal, 1994) before inclusion in the present study and met the criteria as detailed in Table 2.

Considering that most girls with classical RS are profoundly retarded and rarely can perform tasks at a level above a mental age of 1 year, some of the most communicative forme fruste cases in the national Swedish RS complex series, along with some classic, profoundly retarded cases were recruited. The rationale for this design was that a sample of severe and mild

Table 1 **Basic clinical data on the eight RS individuals[a]**

Characteristics	*Classical RS*			*Forme fruste RS*				
	1	2	3	4	5	6	7	8
Normal pre- and perinatal period	+	+	+	+	(+)[b]	+	+	+
Normal development first 6 months	+	+	+	+	(+)[c]	+	+	+
Stagnation onset around (months)	9	14	9	6	18	9	12	10
Regression onset around (months)	18	24	15	20	18	24	20	12
Partial recovery started around (months)	24	30	36	60	48	36	60	?
Seizures	+	+	+	–	–	+	–	+
Seizure onset (years)	8	8	4	–	–	7	–	1
Birth weight (kg)	3.7	3.6	3.6	4.4	3.5	3.3	3.2	3.7
Head circumference at birth (cm)	35	36	34	36.5	36	35.5	33	?
Head circumference current (cm)	54	53	50	55	51.5	50.5	50.5	54
Walks unsupported	+	(+)	–	+	+	+	+	+
Handedness	R	L	?	L	R/L	R	R?	R
Fine motor dyspraxia	+	+	+	+	+	+	+	+
Grasps objects	+	+	–	+	+	+	+	+
Holds objects	(+)	+	+	+	+	+	+	+
Releases objects	+	+	–	+	+	+	+	+
Pincer grasp	–	–	–	+	+	(+)	–	–
Opposition grasp	–	–	–	+	+	+	+	+
Normal vision	+	+	+	+	(+)	(+)	(+)	+
Normal hearing	+	+	+	+	+	+	+	+

[a] + = well documented, clearly holds; (+) = supported by some data, but not completely certain; – = no convincing evidence.
[b] Minor physical anomaly, i.e. cleft palate.
[c] Very 'difficult child' for first 4 years of life.

Table 2 **Applied main and supportive criteria according to Hagberg and Skjeldal (1994) on the eight RS individuals[a]**

Characteristics	*Classical RS*			*Forme fruste RS*				
	1[b]	2[b]	3[c]	4[d]	5[e]	6[d]	7[e]	8[d]
Age (years)	16	22	31	11	17	24	29	36
Main criteria (at least 3 of the following 6)								
A1 Loss of fine motor skill	+	+	+	+	+	–	+	(+)
A2 Loss of nuanced babble/speech	+	+	+	+	+	+	+	+
A3 RS type hand stereotypies	+	+	+	(+)	–	+	+	+
A4 Early onset communication disorder	+	+	+	+	+	+	+	+
A5 Deceleration of head growth	(+)	+	+	–	–	+	(+)	–
A6 Characteristic RS stage development	+	+	+	+	(+)	+	(+)	+
Supportive criteria (at least 6 of the following 11)								
B1 Breathing irregularities	+	+	+	+	+	+	+	+
B2 Bloating or marked air swallowing	–	(+)	–	+	–	(+)	(+)	–
B3 Characteristic teeth grinding	+	+	+	–	+	–	+	+
B4 Gait dyspraxia	+	+	+	+	+	+	+	+
B5 Scoliosis/high kyphosis	+	+	+	(+)	+	+	+	+
B6 Lower limb neurological signs	+	+	+	–	+	+	(+)	(+)
B7 Small, blue, cold feet	+	+	+	–	–	(+)	+	+
B8 EEG sleep pattern characteristic of RS	+	+	+	+?	+?	+	+	+
B9 Unmotivated sudden laughter/screaming	+	+	+	+	+	+	+	+
B10 Impaired delayed nociception	+	+	+	(+)	+	+	+	+
B11 Eye pointing	+	+	(+)	–	–	+	–	(+)

[a] + = well documented, clearly holds; (+) = supported by some data, but not completely certain; – = no convincing evidence.
[b] Classical RS stage III: ambulant.
[c] Classical RS stage IV: non-ambulant.
[d] Forme fruste: no speech; walking.
[e] Forme fruste: speech; walking/running.

cases would offer the best possibility for the study of communication and general cognitive ability.

Procedure

In order to create an optimal setting for the assessment of the participants, primary caregivers and participants stayed at a resource centre (Ågrenska, Göteborg) for children with disabilities in western Sweden. They were seen by a multidisciplinary team consisting of neuropaediatricians, neuropsychiatrists, neuropsychologists, and speech–language pathologists. A multiple case study design was used for the description and differentiation of individual patterns of performance and behaviour.

Assessment methods

Several sources of information such as medical records, parent and staff reports, video recordings, and observations were used for description of the participants' natural history and current functioning. The use of standardized methods was limited by the lack of assessment tools adapted for the unique RS deficiency profile.

The Vineland Adaptive Behavior Scales (Sparrow et al., 1984) were used for the assessment of developmental level of expressive and receptive communicative functions, socialization and motor skills.

An unpublished study (Dahlgren Sandberg, 1997) was used for assessment of communicative and linguistic abilities. The interview was an adaptation for RS subjects of a questionnaire that had been constructed by the first author (ADS) for the purpose of assessment of similar abilities in children with severe cerebral palsy. The interview covered the following areas: early pre-linguistic and linguistic development, communicative modes, current expressive and receptive abilities, communicative function, content and motivation, examples of joint attention and communicative intent. Informants were parents and in some cases personnel who were judged to know the girls well. The current instrument was used because there are no standardized tests for such severely disabled persons in Sweden.

Observation in structured situations was also used to evaluate communicative abilities, specifically verbal comprehension, and social interaction. The sessions were videotaped. Following the direction of the examiner's index finger pointing and attempts at joint attention behaviours were observed. Verbal comprehension at word level was assessed, using two, three or four objects or pictures. The task was to indicate the picture or object that corresponded to the word spoken by the examiner. Standardized tests were tried in those two cases who were judged to function at a communicative and cognitive level advanced enough for the test procedure. The tests used were PPVT–III (Peabody Picture Vocabulary Test III) (Dunn and Dunn, 1997), which assesses verbal comprehension at word level, and TROG (Test of Reception of Grammar) (Bishop, 1989). Since assessments of developmental level may be hazardous in this severely disabled population, the TROG was used in spite of its floor level being 4 years. The reason for doing this was an attempt to try to ascertain the maximum level of functioning.

The examiner was unknown to the participants and informants, and so was the setting. This might have negatively influenced the participants' sense of comfort and subsequent behaviour. However, it is also likely to have decreased the risk of biased findings, owing to prior knowledge on the part of the investigator and to being in a very well-known environment on the part of the RS women.

A DSM-IV checklist for autistic disorder was used and based on informal interviews and described observations. This checklist covers the 12 DSM-IV triad symptoms in detail. By definition, however, according to criterion C, Rett disorder excludes a diagnosis of autistic disorder.

Results

Developmental and communicative levels according to the Vineland Adaptive Behavior Scales

According to the Vineland Scales, the general developmental level varied from 5 to 36 months (Table 3). The highest level for the classic RS cases was 13 months. Three women with forme fruste RS also performed at this level. Two high-functioning forme fruste RS cases were atypical in showing relatively much higher levels of functioning.

There were wide variations concerning communicative development both as regards expressive functions (from under 1 month through 38 months) and verbal comprehension (from 9 through 50 months). In all cases except one, receptive levels surpassed expressive levels as measured on the Vineland Scales. Concerning socialization, scores were higher on interpersonal relationships than on 'play and leisure time' and 'coping skills'. In five out of the eight cases, gross motor skills were better retained than fine motor skills. The opposite was true for the two girls with forme fruste RS who obtained the highest composite scores.

Table 3 **Developmental level in months according to the Vineland Adaptive Behavior Scales**

Case	*Communication*		*Socialization*			*Motor skills*		*Adaptive behaviour composite score*
	Expressive	*Receptive*	*Interpersonal relationships*	*Play, leisure*	*Coping skills*	*Gross*	*Fine*	
Classical RS								
1	3	18	13	8	16	16	7	13
2	10	20	8	<1	<1	24	4	12
3	<1	9	3	<1	–	3	<1	5
Forme fruste RS								
4	10	10	8	<1	<1	22	27	12
5	38	50	35	10	23	29	44	36
6	9	13	14	<1	10	33	13	12
7	23	49	49	15	40	26	32	30
8	1	11	3	<1	<1	24	11	6

Linguistic and communicative development according to structured interview and observation

Tables 4 and 5 summarize results from the structured observations and the interviews.

Development at the time of regression, loss and regain All eight study cases had normal onset of babbling and had developed their first words at onset of regression. They had developed a vocabulary within 'normal limits'. Total loss of speech appeared between 12 and 24 months of age in connection with psychomotor arrest. Two participants in the forme fruste group regained speech at 7 years (case 5) and 8 to 9 years of age (case 7), respectively. Cases 1 and 3, who were among the girls with the largest vocabulary before regression (10 single words), were severely disabled classic RS cases.

Expressive and receptive abilities Only two individuals used oral speech as their predominant expressive mode. Some of the girls vocalized. Most of them demonstrated with bodily or gestural actions their will or, more often, reluctance to participate, e.g. they closed their mouths when not hungry, lay down when they did not want to take further steps or, as the parents said, 'made themselves heavy'. All used eye communication, i.e. 'talked with the eyes' in seeking contact or confirmation, or for example looked at the refrigerator when they were thirsty. Cases 1, 2 and 6 also used 'eye pointing', a conscious act of pointing with the eyes instead of using the index finger, to obtain an object (proto-imperative) or to inform the partner about something (proto-declarative), a way to conduct a conversation. In cases 5 and 7 eye pointing was not demonstrated, since the girls were able to use modes of communication of more symbolic value. The structured observation did not show slow psychomotor responses or dyspraxia as a major limiting factor in expressive abilities.

With the exception of participant 4, who exhibited no difference between expressive and receptive language ability, all the participants had better receptive than expressive linguistic capacity, according to the Vineland Scales. Most participants were judged by their parents to understand only key words, one of them only her own name. Simple coherent speech with visual support was clearly understood by three of the participants. Only two girls with expressive speech were believed also to understand at least simple sentences without visual support.

The communicative aim that was most easy to understand and to interpret was expression of primary needs (Table 5). Six of the cases could also express basic feelings such as happiness and sadness in an intelligible way, while cases 5 and 7 demonstrated more differentiated feelings. Only case

Table 4 Linguistic and communicative functions according to the structured interview and direct observations[a]

Case	Developmental level before regression		Regain of speech	Communicative functions					Predominant communicative form	Intentional gestures			Attitude to communication
	Babbling	No. of words		Joint attention	Communicative intent	Expression of primary needs	Need for social conmtact	Sharing information		Giving at request	Pointing: whole hand	index finger	
Classical RS													
1	+	10	–	+	–/+	–/+	+	–	Humming, eyes, face, body	–	+	–	Positive
2	+	2	–	–	–	–/+	+	–	Humming, eyes, smiling, body	–	–	–	Postive
3	?	10	–	–/+	–	–	–	–	Whining, eyes, face	Not possible	–	–	Rare demonstration of communicative will
Forme fruste RS													
4	?	6	–	–	–	+	–/+	–	Pictures, whining, gestures, face, body	–	–	–	At times rather positive
5	+	2	At 7 years, sentences of 2–5 words, sometimes function words	+	+	+	+	+	Speech, manual signs	+	+	+	Positive
6	+	3	The word 'yes'	–/+	–	–	+	–	Manual signs, gestures, eyes, face, body	–/+	+	–	Positive
7	?	>10, 3-word sentences	At 8–9 years, sentences of 1–2 words	+	+	+	+	+	Speech, pictures, gestures, face, body	+	+	+	At times rather positive
8	?	3	–	–	–	+	+	–	Whining, eyes, body	–	–	–	Hardly any will

[a] + = present; – = absent; –/+ = mostly absent, occasionally present; (+) = demonstrated once during observation.

Table 5 **Understanding of speech and intelligibility in the eight RS individuals[a]**

Case	*Understanding of speech*	*Intelligibility or possibility to understand/interpret*		
		Primary needs	*Drive for preferred activities*	*Expression of feelings*
Classical RS				
1	Key words, simple sentences with visual support	Primary care-givers only	–	Happy/sad
2	Key words, simple sentences with situational and visual support	With visual support	–	Happy/sad
3	Doubtful if any	–	–	Happy/sad
Forme fruste RS				
4	Very limited with situational support	+	+	Happy/sad
5	Consistent with mental age	+	+	+
6	Key words, simple sentences about well-known things	–	–/+	Happy/sad
7	Key words, simple sentences	+	–/+	+
8	Own name	With visual support	–	Happy/sad

[a] + = yes; – = no; – /+= mostly no, occasionally yes.

5 could make herself clearly understood when wanting to perform preferred activities. Otherwise, the individuals moved to the desired object, and stayed there until the parent interpreted their behaviour and acted on that interpretation.

Joint attention and intentional communication Joint attention, considered to be a prerequisite for intentional communication, was demonstrated clearly by only three of the participants, the two high-functioning girls of forme fruste RS variant, and one with classic RS. In another case (6), the mother gave anecdotal evidence of one occasion of joint attention. The remaining participants did not show any signs of joint attention behaviour. Consequently, communicative intention differed among participants. Cases 2, 3, 4, 6 and 8 showed no signs of intentional communication. They were dependent on their caregivers' interpretation of their non-verbal expressions.

Autistic features Case 4 and case 8 with forme fruste RS met the DSM-IV symptom criteria for autistic disorder as shown in Table 6. The scoring was based on medical records, video recordings, and observation during

Table 6 **Symptoms of DSM-IV autistic disorder (American Psychiatric Association, 1993) in the eight RS individuals[a]**

Criterion[b]	*Classical RS*			*Forme fruste RS*				
	1	2	3	4	5	6	7	8
A1 *Social interaction impairment*								
(a) Impaired interaction regulation	–	–	–	+	–	+	–	+
(b) Failure to develop peer relations	–	–	–	–	–	–	–	–
(c) Lack of sharing	–	–	–	+	–	–	–	+
(d) Lack of reciprocity	–	+	–	+	–	–	–	+
A2 *Communication impairment*								
(a) Delay/lack of spoken language	+	+	+	+	–	+	–	+
(b) Impaired conversational skills	–	–	–	–	–	–	–	–
(c) Stereotyped language	–	–	–	–	+	–	+	–
(d) Lack of varied play	–	+	–	+	–	–	–	+
A3 *Behavioural impairment*								
(a) Stereotyped and restricted interests	–	–	–	+	+	+	–	+
(b) Non-functional routines/rituals	–	–	–	–	–	–	–	–
(c) Motor stereotypies	+	+	+	+	+	+	+	+
(d) Preoccupation with object parts	–	–	–	–	–	–	–	–
B Met criteria for RS[c]	+	+	+	+	+	+	+	+

[a] + = well documented, clearly holds; – = no convincing evidence or not relevant.
[b] For details see text of DSM-IV.
[c] Excludes a diagnosis of autistic disorder.

assessments. These two girls also scored very low on the Vineland subscales for interpersonal relationships and coping skills.

Discussion

General developmental level, as indicated by Vineland composite scores, suggested overall very low functioning in RS. Three distinct developmental levels could be identified. Two girls performed at a developmental level of 5 and 6 months, four girls at 12 and 13 and two at 30 and 36 months, respectively. Interestingly, there was no difference in distribution in this respect across classical RS and milder forme fruste RS cases. However, two relatively high-functioning forme fruste RS cases stood out as atypical, not only in comparison with the classical RS girls, but also within the forme fruste group. These two individuals regained speech during late childhood and showed relatively good social abilities. Thus, general developmental level in this study did not seem to conform in any clear way to clinical attributes.

It could be argued that any conclusion based on a single assessment of cognitive level is a hazardous enterprise in individuals with such a restricted repertoire, in respect of both motor performance and communication. However, independent evaluation based on the detailed structured interview on communication and the video recorded assessment of communicative and social interaction behaviour provided a wealth of corroborative information consistent with the Vineland data.

Contrary to the retrospective results of Tams-Little and Holdgrafer (1996), all participants in this study showed normal onset of babbling and appearance of first words until onset of developmental arrest which, according to parental reports and medical records, occurred from 12 to 24 months of age. The speech used at developmental arrest, varying from two to somewhat more than 10 words, was subsequently lost in all cases. In two forme fruste cases, spoken language was regained only at 7–9 years of age. One of these two girls had used appropriate three-word sentences at regression onset at 20 months of age. At age 29 years, she performed at the same linguistic level that she had acquired at the onset of regression, i.e. mostly single-word utterances and some two- or three-word combinations. The other forme fruste girl with some spoken language had acquired two single words at developmental arrest at 18 months of age. At 7 years of age she regained spoken language. At assessment, this girl used fully formulated short sentences, sometimes omitting function words and using incorrect word order, roughly corresponding to her mental age of 36 months and to her results on the PPTV–III and the TROG. Speech intelligibility was very low at times. The results suggest that, to be able to use speech after regression, subjects with forme fruste RS must function on a developmental level of 30–36 months of age.

At onset of regression, there were no obvious differences in linguistic ability between classical and forme fruste RS. Two of the classical RS cases at that time outperformed all but one of the other cases, using 10 single words, respectively. These words were lost and never regained. Thus, in this small sample, linguistic level at developmental arrest did not seem to predict later speech ability. These results do not support the findings of von Tetzchner et al. (1996) who found that the number of words at regression predicted the number of words later regained.

Non-verbal modes of communication (eye communication, and bodily and gestural actions) were mostly used to express reluctance not pleasure. Eye pointing in a clear communicative sense was found only in three of the girls. In two cases (5 and 7), eye pointing was not demonstrated, probably because the girls were able to use language for communication. In those six individuals who functioned at a developmental level of 12 months or more, intentional gestural communication could have been expected. However,

clear evidence of gestures such as pointing and giving, considered important markers of development from a pre-linguistic to linguistic level, was found only in those who were already at a linguistic level. Thus, the absence of these motor activities did not seem to be explained by slow psychomotor response or by dyspraxia, but rather by a generally low level of communicative development. The limited use of gestural intentional communication corroborates findings from other studies (e.g. Tams-Little and Holdgrafer, 1996).

Comprehension of spoken language was usually restricted to own name, key words or simple sentences with visual or situational support (equally distributed among the classical and the forme fruste cases). However, the two girls with forme fruste RS previously discussed stood out as atypical. Their receptive abilities were judged to be in accordance with mental age, in one instance, and restricted to simple sentences expressed orally in the other. The receptive language of one individual (case 3) was difficult to evaluate because of her limited possibility for response owing to extreme physical disability.

Thus, in seven of the eight cases in this study, receptive ability exceeded expressive ability, as assessed by the Vineland Scales. The eighth girl performed equally well receptively and expressively. These results contrast with the findings of Woodyatt and Ozanne (1992a; 1992b; 1993) and von Tetzchner (1997) who found equally low comprehension and level of spoken language. However, they do accord with those of Zappella (1997).

Joint attention behaviours were demonstrated by only three of the participants, the two most high-functioning cases of forme fruste RS and one classical RS participant (case 1). According to developmental level, joint attention could have been expected in three further cases, the participants with a Vineland composite score of 12 months. In two cases (4 and 8) the combination of low developmental level and severe autistic social impairment probably accounted for the lack of joint attention. Since joint attention is considered a prerequisite for intentional communication, communicative intent, not surprisingly, was found only in the two most high-functioning girls. There was also some evidence of intentionality in the third girl who showed joint attention. In spite of little demonstration of communicative intent, most of the participants (cases 1, 2, 5, 6 and 7) showed social interest, as evidenced by social smiling and looking at the communicating partner. All participants also scored higher on the Vineland subscale 'interpersonal relationships' than on 'play and leisure time' and 'coping skills', which might demonstrate the interest in social contact that has been observed in other studies on RS. However, the low results on 'play and leisure time' might also reflect difficulties with make-believe activities, cooperation in interaction games, motor planning and motor performance.

The obvious social withdrawal and aloofness in two cases impeded communicative responses. Again, such abilities were difficult to assess in one case (3).

Two forme fruste RS cases met full DSM-IV symptomatic criteria for autistic disorder as demonstrated in Table 6. This finding is in conjunction with earlier reports of overlapping autistic and RS features among atypical RS cases (Zappella, 1994; 1997; Zappella et al., 1998). These girls also scored very low on the Vineland subscales for interpersonal relationships and coping skills. Two individuals with classical RS (cases 2 and 3) scored equally low on the same Vineland subscales, but presented only a few autistic features. In these cases, the major factors accounting for low social and coping skills seem to be the extreme form of RS physical disability (case 3), and disabling hyperventilation attacks (case 2). The present and previous findings of RS and autistic comorbidity in some adult forme fruste RS cases may be taken to invalidate the DSM-IV criterion C, which posits that a diagnosis of RS excludes a diagnosis of autistic disorder. Also, there has been critique (e.g. Gillberg, 1994) of the DSM-IV inclusion of RS as one particular named variant of the autism spectrum disorders (or 'pervasive developmental disorders').

In summary, this study presents eight RS complex girls and women, all with severe cognitive difficulties, but with varying degrees of communicative abilities not corresponding to their cognitive functioning and not following expected developmental patterns. Communicative functions at developmental arrest did not seem to predict later language ability, or type of overall clinical presentation. Expressive and receptive abilities were at low levels, as were joint attention and communicative intent. Means of communication varied according to individual ability, with more expression of symbolic quality in the most able girls. Eye pointing, as distinct from eye communication, was demonstrated only in a minority of cases. Even when taking account of general developmental level, the girls and women in this study showed communicative and linguistic difficulties that seem to be specific to RS psychopathology.

The two cases with a clear symptomatic picture of autistic disorder contrasted with the other participants in that they did not show interest in social interaction.

Since most of the girls seemed to function at a pre-intentional level, with little evidence of joint attention or use of gestural intentional communication, intervention should probably focus on enhancing such actions in communication. Another area of possible intervention would be the observation, maintenance and development of the communicative actions spontaneously used by the girls, such as eye pointing, gestures, pictures, manual signs and, in some cases, spoken language.

A problem in the present study was to find assessment methods adapted to the low developmental level, relatively high chronological age of the participants and extraordinary impairment characteristics of RS girls. The small number of cases is also a limitation. Consequently, the data must be treated with caution. Studies with large samples are not common, though, in this field. Only the Zappella et al. (1998) and von Tetzchner (1997) studies included a relatively large number of cases, 30 and 42 respectively. Development of methods for the study of this group of very disabled individuals should be a priority in future investigations of the cognitive, communicative and linguistic functioning and dysfunctioning in RS complex.

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