The Relationship of Cumulative Motor Asymmetries to Scoliosis in Rett Syndrome

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Key Words: body alignment • movement disorders

Objectives. Interrelationships between Rett syndrome scoliosis and symmetric and asymmetric motor pull, ambulation, and advancement of age were investigated in order to provide a treatment rationale for slowing the progression of scoliosis.

Method. Questionnaires (N = 262) completed by International Rett Syndrome Association families were analyzed with logistic regression, odds ratio, Kruskal-Wallis one-way, and Fisher's exact test analyses.

Results. Rett syndrome scoliosis was found to be significantly related to orthopedic risk factors of asymmetric movements and positions (odds ratio = 4.5, p < .001). The odds ratio for asymmetric upper-body positioning (4.4), nonambulation (3.1), and age advancement (4.1) indicated that each was a significant predictor of scoliosis by logistic regression analysis. Univariate Kruskal-Wallis one-way analysis identified a significant relationship between scoliosis and asymmetric higher shoulder positioning (p < .001) and bodyside movements (p = .03). Hemisyndrome aspects of Rett syndrome were identified as the increased prevalence of symptoms in either right or left bodyside.

Conclusion. A significant relationship was found between the prevalence of Rett syndrome scoliosis and orthopedic risk factors. These findings suggest a treatment approach for Rett syndrome scoliosis that focuses on balancing bilateral muscular pull.

Rett syndrome is a debilitating disorder (Rett, 1977) of unknown etiology (Hagberg, 1995) that has been reported to be the result of a neurodevelopmental arrest (Armstrong, 1995; Armstrong, Trivedi, & Glaze, 1994; Hanefeld, Hagberg, & Percy, 1995), with an estimated prevalence of 1 case per 10,000 live-born girls (Hagberg & Witt-Engerstrom, 1987). Rett syndrome was characterized initially as a progressive syndrome of autism, dementia, ataxia, and loss of purposeful hand use (Hagberg, Aicardi, Dias, & Ramos, 1983). Hard signs of this disorder appear between 6 and 18 months of age as regression in motor development; loss of purposeful hand movements; and onset of involuntary, stereotyped arm and hand movements (Treichan & Naïdu, 1988), mostly at midline (Hagberg, 1993; Nomura & Segawa, 1990), that disrupt both manipulative and gross motor skills (Hanks, 1990). If the ability to ambulate has been acquired, gait apraxia and truncal ataxia are evident (Hanks, 1990; Van Acker, 1991). There is an apparent progressive reduction in muscle mass, beginning at approximately 5 years of age (Hagberg et al., 1983; Naïdu, Murphy, Moscr, & Rett, 1986). A hemisyndrome symp-
Scoliosis is the primary musculoskeletal, orthopedic complication of Rett syndrome (Hennessy & Haas, 1988). Rett syndrome scoliosis is reported to be of a neurologic type (Stokland, Lidstrom, & Hagberg, 1993) and associated with the neurologic symptoms of dystonia of the trunk (FitzGerald, Jankovic, Glaze, Schultz, & Percy, 1990), spasticity, wasting, and incoordination (Harrison & Webb, 1990). It develops initially with alterations in axial muscle tone, with hypertonicity on one side of the spinal curve and hypotonicity on the other side (Van Acker, 1991). A slowing down of righting and equilibrium reactions is seen in the development of scoliosis (Budden, 1995). Scoliosis progresses with age (Guidera, Borrelli, Raney, Thompson-Rangel, & Ogden, 1991; Harrison & Webb, 1990; Stokland et al., 1993), along with muscle weakness and hypotonia (Stokland et al., 1993; Witt-Engerstrom, 1990), and becomes fixed in advanced stages of Rett syndrome (Hanks, 1990). Spinal rigidity is reported to be "voluntary" as a compensatory mechanism for loss of truncal stability (Hanks, 1990). Degrees of severity in spinal curve and spinal curve patterns vary widely (Hennessy & Haas, 1988) (see Figure 1). Correspondingly, there is variety in asymmetric extremity and truncal body positions, stereotypic arm and hand movements, and limb wasting (Hagberg, 1993). The development of nonfunctional stereotypic arm and hand movements precedes the development of scoliosis (Trevarhan & Naidu, 1988). Typically, these jerky, nonfunctional movements of trunk and limbs are seen by 3 years of age (Van Acker, 1991) in Stage II of the disease (Trevarhan & Naidu, 1988).

Scoliosis becomes more evident when the child approaches adolescence (Van Acker, 1991). In Stage III of the disease, at 2 to 10 years of age, early scoliosis can appear; progressive scoliosis appears at 10 years of age and older in Stage IV (Trevarhan & Naidu, 1988). Cases of severe scoliosis are more common in girls with more severe Rett syndrome (Harrison & Webb, 1990; Witt-Engerstrom, 1990) who are nonambulatory (Naidu et al., 1986). The most severe curves are present in girls who have never walked (Harrison & Webb, 1990). Scoliosis is reported to be treatable with spinal fusion when it progresses beyond 40° Cobb angle, a standard method of measuring spinal curve (Harrison & Webb, 1990). Stokland et al. (1993) recommended meticulous spinal curve monitoring of any-aged girls with Rett syndrome when early hypotonia and motor impairment are present. Scoliosis is a major factor of morbidity (Witt-Engerstrom, 1990) in the form of painful respiratory compromise (Keim & Hensinger, 1989) that leads to pneumonia (Glaze, 1995).

In addition, persons with Rett syndrome have demonstrated hemisindrome symptoms, that is, a tendency for symptoms to be either right or left sided. Budden (1995) found that of the 43% out of 60 girls with Rett syndrome who retained ambulatory skills, 62% showed right-sided lower-extremity weakness, and 38% showed left-sided lower-extremity weakness. The muscular imbalance of the hemisindrome symptoms may lead to the development of the orthopedic complication of scoliosis and to the scoliotic curve patterns that have been reported to be more frequently to the right than to the left (Guidota et al., 1991).

Maintaining mobility in daily activities through ambulation is an important goal of therapy (Hanks, 1986). Therapeutic intervention with active muscular exercise has been shown to maintain and regain ambulatory skills and possibly stabilize and resolve scoliosis. In a retrospective study of 23 persons with Rett syndrome from 2 to 21 years of age, 15 ambulated initially, and 11 maintained ambulation skills (Hanks, 1990). Of those who lost ability to ambulate, the 4 girls had regained it after therapeutic intervention; length of treatment was not reported. Clare (1986) reported that a rigorous physical exercise program of 8 months duration helped a child with Rett syndrome who initially was unable to bear weight to achieve supported ambulation. The program also may have helped to stabilize the child’s scoliosis; specific degrees of scoliotic curve were not given. Another child in the same study was able to regain the ability to stand after intensive hydrotherapy and many repetitions of amбула-
of these girls required spinal fusion, and all received some clinical and radiological resolution (Budden, 1995). None
syndrome neuromuscular deficits: "Why are neuropatholog­
[52x550]al abnormalities seen peripherally, including scoliosis, have
been reported to be neurologic in origin (Badr, Witt-Engers­
gerstrom, & Hagberg, 1987; Harrison & Webb, 1990),
but the site of origin of these CNS symptoms has not
been identified within the brain (Haas & Love, 1988; 
Hagberg et al., 1983; Hennessy & Haas, 1988). Haas
(1988) asked the all-important question regarding Rett
syndrome symptoms result in CNS orthopedic
asymmetries through axial, truncal muscular pull of repeti
tive motion and position (see Figure 2). Thus, the etiology
of the neuropathological area of deficit may involve only
that region of the brain causing the stereotypies and
hemisyndrome symptoms, explaining the limited brain
neuropathologic changes along with the vast number and
variety of CNS orthopedic changes noted with this syn­
drome. We developed a hypothesis that the development
of scoliosis is orthopedic in origin and is the result of the
cumulative, muscular pull of one or more motor asymme­
tries resulting from hemisyndrome symptoms, stereotypic
movements, or other neurologic effects. These muscle
asymmetries could result in scoliosis, followed by subse­
cutive postural positioning changes in an attempt to regain
midline orientation or positioning changes that could lead
to the development of scoliosis (see Figure 2). The wide
variety of asymmetries in movements and positions ob­
served clinically could produce the variety of spinal curve
patterns that are characteristic of Rett syndrome (see
Figure 1). As the child with Ret( syndrome ages, the
cumulative muscular asymmetries would continue to give
an unbalanced, asymmetric pull, leading to the clinically
observed increases in severity in scoliosis with age. Sym­
metry in stereotypic movements at midline and symmet­
crical positions, which could result in a more bilaterally
symmetric pull, could result in lower prevalence and
severity of scoliosis in persons with Rett syndrome.
This study was designed to evaluate risk factors of
abnormal upper-body positions and movements as well
as ambulatory status in relation to the prevalence of scoli­
osis in persons with Rett syndrome. A better under­
standing of the relationship of these risk factors to the
development of scoliosis could provide a rationale for an
Figure 2. Schematic of the proposed etiology of Rett syn­
drome scoliosis.

occupational therapy treatment approach to slow the
progression of scoliosis and to maintain the functional
independence of the child with Rett syndrome.

Method
Participants
Participants were recruited from the International Rett
Syndrome Association (IRSA) database on the basis of the
following criteria: (a) confirmed diagnosis of Rett syn­
drome, (b) U.S. residency, and (c) permission given to be
contacted for research purposes. Persons meeting these
criteria were further identified by age (within a range of
2.5 years to 30 years) in order to ensure representation
from four age groups: < 3 years, 3 to 5 years, 6 to 9 years,
and > 10 years. These age groupings approximate the
four progressive stages of Rett syndrome. The number of
eligible participants identified for each age group was 8
(< 3 years), 63 (3–5 years), 130 (6–9 years), and 676 (> 10
years). All 71 participants in the two youngest age
groups were included in the study, and 129 of 130 eligi­
ble participants were included in the 6 to 9 years age
group. To represent the > 10 years age group, participants
were randomly selected until a total of 200 was secured
(see Table 1).

Instrument
A questionnaire was designed to obtain the following
information: the participant’s birth date and question­
aire completion date, presence of abnormal hand or arm
movements (Question 1), laterality or symmetry in hand
and arm movements (Question 2), laterality or symmetry
in arm and shoulder positions (Questions 3 and 4),
descriptive scoliosis information of the direction and
degree of curve (Questions 5 and 6), and current ambula­
tion status (Question 7).
The participant’s age was calculated from the birth
date and the questionnaire completion date. Question 1,
the identifying diagnostic marker of Rett syndrome, was
scored as either yes or no. Question 2 (designated as
hand–arm) identified the symmetry of hand–arm move­
ments and was scored as right (asymmetric), left (asym­
metric), or both (symmetric). Question 3 (designated as
bodyside) identified the symmetry of movements in rela-
Table 1

<table>
<thead>
<tr>
<th>Age Group (Years)</th>
<th>Number in IRSA Database</th>
<th>Surveys Sent</th>
<th>Surveys Returned and Analyzed</th>
<th>Ambulatory Status</th>
<th>Scoliosis Status</th>
</tr>
</thead>
<tbody>
<tr>
<td>I (&lt;3)</td>
<td>8</td>
<td>8</td>
<td>4 (50)</td>
<td>2 (50)</td>
<td>1 (25)</td>
</tr>
<tr>
<td>II (3-5)</td>
<td>63</td>
<td>63</td>
<td>32 (51)</td>
<td>15 (47)</td>
<td>8 (25)</td>
</tr>
<tr>
<td>III (6-9)</td>
<td>130</td>
<td>129</td>
<td>64 (50)</td>
<td>35 (55)</td>
<td>25 (39)</td>
</tr>
<tr>
<td>IV (≥10)</td>
<td>676</td>
<td>200</td>
<td>128 (64)</td>
<td>66 (52)</td>
<td>100 (78)</td>
</tr>
<tr>
<td>Total</td>
<td>877</td>
<td>400</td>
<td>228 (57)</td>
<td>118 (52)</td>
<td>134 (59)</td>
</tr>
</tbody>
</table>

Note. n = 228. IRSA = International Rett Syndrome Association.

Relationship to body midline and was scored as right (asymmetric), left (asymmetric), at midline (symmetric), or one sided (asymmetric). By combining hand–arm and body side movement scores (Questions 2 and 3), a combined movement category was designated and scored as symmetric if both categories were symmetric and as asymmetric if either hand–arm or body side movements were asymmetric. Question 4 (designated as higher shoulder) was scored as right, left, or symmetric. To indicate asymmetry in movement (hand–arm, body side) and positioning (higher shoulder), a total asymmetry category was designated and scored as symmetric or, if asymmetry was present, as asymmetric right or asymmetric left. When scoliosis was present and both left and right asymmetries were seen, the asymmetry result was not used in the directional analysis of the scoliosis curve involving total asymmetry. Questions 5 and 6 were scored yes or no; right, left, or both; and degree of scoliosis if known (for Question 6). Question 7 was scored as yes or no. An I don't know option was included for each question to assess the validity of family members’ responses.

Procedure and Scoring

IRSA identified potential participants and mailed the questionnaire to their family members in order to maintain confidentiality. The anonymous, completed surveys were returned to the investigators. Responses were scored independently by two investigators and entered into the R:Base database. These investigators collaborated later to correct and resolve discordant scores.

Data Analysis

Distributions, including the incidence of scoliosis within age groups, ambulation status, and symmetry of positioning and movements, were obtained from the R:Base database. The Kruskal–Wallis one-way analysis was used to address the questions of whether participants with asymmetric positioning or movements or nonambulatory status were more likely to have scoliosis. Univariate analyses of categorical variables were carried out with a chi-square statistic, except in cases where the values in the cells were too small (expected > 5), in which case the Fisher’s exact test was used. These tests were used because the data were nonparametric; that is, they were not normally distributed. The odds ratio was obtained for the variables of age, ambulation, and asymmetry of positioning and movement. The odds ratio is the ratio of the odds of getting the disease (scoliosis) with and without the exposure variable (e.g., age, ambulation, asymmetry of positioning, asymmetry of movement) (Rothman, 1986). BMDP Statistical Software was used to compute the logistic regression, with scoliosis as the outcome and age, ambulation, and asymmetry of positioning or movement as possible predictors. Logistic regression has been used extensively in epidemiologic research to establish the risk of disease (scoliosis) associated with a set of exposures (risk factors) (Portney & Watkins, 1993). The confidence intervals reported for the logistic regression results are the lower and upper limits of the odds ratios, with 95% confidence that the odds ratio of the entire population being sampled will be within this interval (Hosmer & Lemeshow, 1989).

Results

Of 400 questionnaires mailed, 262 were completed and returned (66%). Of these, 32 were omitted because the responses to Question 3 indicated an ambiguous interpretation of the symmetry of movement. An additional 2 questionnaires were omitted because the participant reportedly had no abnormal hand or arm movement (Question 1). The total number of questionnaires submitted to statistical analysis, therefore, was 228 (57%). The number of questionnaires completed per age group is shown in Table 1 (range = 50%-64%).

By comparing the prevalence of scoliosis with that of symmetric and asymmetric movements and positions (the total asymmetry, including any asymmetric movement and position), we found that scoliosis was associated with

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2Release 7.0 is distributed by BMDP Statistical Software, Inc., 12121 Wilshire Boulevard, Suite 300, Los Angeles, California 90025.
asymmetry ($n = 83$) compared with symmetry ($n = 24$) and that no scoliosis was associated with symmetry ($n = 65$) compared with asymmetry ($n = 48$). Of those participants that had asymmetric movements and positions, $3.5$ times more had scoliosis than did not. Of those participants that had symmetric movements and positions, $1.4$ times more did not have scoliosis than did. This univariate analysis of total asymmetry and the prevalence of scoliosis gave a significant Fisher's exact $p < .001$ and an effect of odds ratio $= 4.5$.

The logistic regression analysis of age as the predictor of scoliosis was carried out by two different ways of grouping the questionnaires. The questionnaires were grouped into four age groups that approximated developmental stages of Rett syndrome. However, because the youngest age group ($< 3$ years) had only four participants, we also divided the questionnaires into age quartiles ($0$-$7.25$ years, $7.25$-$10.89$ years, $10.89$-$17.2$ years, $> 17.2$ years) to check whether the small sample size of the youngest age group had affected the results. Logistic regression analysis of age by quartiles gave lower standard errors in the age analysis than did the age group data, which may be due to the small sample size of the first age group. Although the confidence intervals of age by quartiles was high, indicating a large standard error, this does not indicate that they are not significant. We would be $95\%$ confident that persons with Rett syndrome would be between $4.2$ and $33$ times as likely to have scoliosis at $> 17.2$ years of age if it were possible to sample the entire population. The odds ratio was higher with the other age groups (see Table 2). The results of logistic regression analysis of abnormal asymmetric upper-body positioning (odds ratio $= 4.4$, $p < .001$) and inability to ambulate (odds ratio $= 3.1$, $p < .01$) using age groups were the same as the results using the quartile age groups (see Table 3).

Abnormal asymmetric upper-body positioning and inability to ambulate were significant predictors of scoliosis (see Table 2). When the combined hand-arm and bodyside movement scores were entered into the initial regression model along with the predictor variables of age, ambulation, and positioning, the analysis did not retain the combined movement scores as a significant predictor of scoliosis, although univariate analysis of bodyside scores were significant ($p = .03$).

In the total sample ($N = 228$), the prevalence of scoliosis was $59\%$. Using univariate analysis, significant odds ratios to predict scoliosis were obtained with the risk factors of combined movement ($2.1$); nonambulation ($2.4$); the total asymmetry category, including any asymmetric movement or position ($4.8$); and higher shoulder ($6.1$). This odds ratio of the higher shoulder indicates that participants with a higher shoulder asymmetry were $6.1$ times more likely to have scoliosis than a participant without higher shoulder asymmetry. Because the odds ratio is a measure of the magnitude of the effect of the risk factor, the higher shoulder category has a larger effect than do the other three categories.

Approximately $50\%$ of the participants in each of the four age groups were reported to be ambulatory. The data were further analyzed for the effect of movement and positioning on scoliosis by dividing the sample into participants who were ambulatory and participants who were nonambulatory (see Table 3). In participants who were ambulatory, asymmetric hand-arm ($p = .04$) and combined bodyside and hand-arm movements ($p = .015$) had a significant relationship to the prevalence of scoliosis by the Fisher's exact test. In the ambulatory group, the relationship of asymmetric bodyside movement to the prevalence of scoliosis gave an exact $p$ of $0.08$. The odds ratio for all of the movement categories of participants who were ambulatory were greater than or equal to $2.5$. In participants who were nonambulatory, the relationship of asymmetric bodyside movement to the prevalence of scoliosis gave an odds ratio of $3.0$ and an exact $p$ of $0.12$. The odds ratios and Fisher's exact $p$ values were not significant for the reported hand-arm or combined movement asymmetries in these participants. The relationship of the reported higher shoulder positioning asymmetry to the occurrence of scoliosis was significant with the Fisher's exact test and the odds ratios for both participants who were ambulatory (odds ratio $= 6.2$, $p < .001$) and participants who were nonambulatory (odds ratio $= 6.9$, $p < .001$) (see Table 3).

The hemisyndrome symptoms indicated a right predominance. The direction of the spinal curve favored

Table 2
Logistic Regression Analysis of the Prevalence of Scoliosis in Girls With Rett Syndrome in Relationship to Asymmetric Positioning Parameters, Ambulatory Status, and Age

<table>
<thead>
<tr>
<th>Outcome</th>
<th>Risk Factors</th>
<th>Odds Ratio</th>
<th>Confidence Interval$^2$</th>
<th>$P$</th>
</tr>
</thead>
<tbody>
<tr>
<td>Scoliosis</td>
<td>Age (years)</td>
<td>2.5</td>
<td>1.01-6.04</td>
<td>&lt; .5</td>
</tr>
<tr>
<td></td>
<td>7.2-10.9</td>
<td>2.5</td>
<td>1.01-6.04</td>
<td>&lt; .5</td>
</tr>
<tr>
<td></td>
<td>10.9-17.2</td>
<td>8.9</td>
<td>3.3-25.8</td>
<td>&lt; .001</td>
</tr>
<tr>
<td></td>
<td>&gt; 17.2</td>
<td>11.8</td>
<td>4.2-33.1</td>
<td>&lt; .001</td>
</tr>
<tr>
<td></td>
<td>Asymmetric higher shoulder</td>
<td>4.5</td>
<td>2.1-9.5</td>
<td>&lt; .001</td>
</tr>
<tr>
<td></td>
<td>Nonambulation</td>
<td>3.0</td>
<td>1.5-6.3</td>
<td>&lt; .01</td>
</tr>
</tbody>
</table>

Note: Asymmetric movement parameters, although significant in univariate analyses, were not retained as significant independent predictors by logistic regression analysis.

$^2$The confidence interval, calculated from the standard error, indicates significance if it does not include $1$. March 1998, Volume 52, Number 3
right (51%) over left (34%) and S-shaped curve (15%). We found more participants with right total asymmetry (n = 51) than left (n = 43). Right scoliosis corresponded more with right asymmetry in either positions or movements (n = 21) than left asymmetry in either positions or movements (n = 14). Left scoliosis corresponded more with left asymmetry (n = 16) than right asymmetry (n = 11) (odds ratio = 2.2, p = .12). Right higher shoulder corresponded more with right scoliosis (n = 19) than left (n = 11). Left higher shoulder corresponded more with left scoliosis (n = 16) than right (n = 13) (odds ratio = 2.1, p = .12). However, these directional analyses were not significant in this limited sample size. Participants with S-shaped spinal curves were not included in this directional analysis.

There were 107 participants with asymmetric movements and positions (the total asymmetry category) and 113 with symmetric movements and positions. One hundred ninety-seven had symmetric hand–arm movements, and only 15 had right and 13 had left asymmetric hand–arm movements. There were 194 symmetric body-side movements at midline and only 16 right and 16 left of midline. One hundred seventy had combined symmetric movement scores, and 51 had asymmetric combined movement scores.

The Kruskal-Wallis one-way analysis of asymmetry of position and movement compared with the prevalence of scoliosis revealed that the presence of one shoulder being higher was significantly related to those participants who had scoliosis (p < .001). Bodyside movements were also significant (p = .03) but not hand–arm movements (p = .24). The odds ratio of the same higher shoulder compared with the same direction of the scoliosis curve was 2.1. However, this was not significant with an exact p of .16 for this limited sample size.

The reported prevalence of Rett syndrome scoliosis increased dramatically with age from 25% in the youngest two age groups to 78% in the oldest age group (see Table 1). Of the 134 participants with scoliosis, the degree of spinal curvature was reported for 68%. There were no scoliosis curves exceeding 39° in the youngest two age groups. In the 6 to 9 years age group, 27% had curves greater than 39°. There were 51% reported curves greater than 39° in the oldest age group.

### Table 3

<table>
<thead>
<tr>
<th>Symmetry Category</th>
<th>Ambulatory Odds Ratio</th>
<th>p</th>
<th>Nonambulatory Odds Ratio</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Arm–hand movement</td>
<td>3.8</td>
<td>&lt;.04</td>
<td>0.8</td>
<td>&lt;.01</td>
</tr>
<tr>
<td>Bodyside movement</td>
<td>2.5</td>
<td>&lt;.08</td>
<td>3.0</td>
<td>&lt;.12</td>
</tr>
<tr>
<td>Combined movement</td>
<td>3.2</td>
<td>&lt;.015</td>
<td>1.8</td>
<td>&lt;.24</td>
</tr>
<tr>
<td>Higher shoulder position</td>
<td>6.2</td>
<td>&lt;.001</td>
<td>6.9</td>
<td>&lt;.001</td>
</tr>
</tbody>
</table>

### Discussion

Rett syndrome scoliosis may be due primarily to continued pull of orthopedic asymmetries over time rather than being a neurologic form of scoliosis. The cumulative effect of one-sided muscular activity may result in a hypertrophic development on the side of the spinal hyper tonicity and a hypotrophic muscular development on the hypotonic side of the spine with its hypotrophic muscular development. We reasoned that asymmetric muscular pull causes Rett syndrome scoliosis and not that scoliosis causes the asymmetric muscle imbalance because the stereotypic movements and the muscle strength imbalances precede structural changes in the spine.

Asymmetries in strength of the proximal, paraspinal, and axial muscles may represent a more dominant factor in the development of Rett syndrome scoliosis than distal hand muscle asymmetries. Our results (to our knowledge not previously reported) indicate that both the size of the effect and the significance was greater for the prevalence of scoliosis when there was higher shoulder asymmetry with proximal truncal muscles than when distal hand–arm asymmetry was present (see Table 3). Intensive therapeutic intervention to balance the bilateral muscular pull of Rett syndrome scoliosis would result theoretically in promoting a balanced bilateral muscular pull to both sides of the body. Abnormal, asymmetric bodyside movements may be a supporting factor in predicting scoliosis. Bodyside movements that have proximal components of muscle origins may represent a stronger influence than distal hand–arm musculature but a weaker influence than axial and shoulder musculature components. Our finding that Rett syndrome scoliosis is highly correlated with proximal asymmetric muscles suggest that occupational therapy practitioners work closely with physical therapists to incorporate a more bilaterally symmetrical muscular balance through proper sitting and lying positions and maximal involvement of the child in self-care tasks.

Higher shoulder asymmetry was a significant predictor of scoliosis; however, the direction of the curve did not significantly correspond with the direction of the higher shoulder asymmetry. Because this is not a longitudinal study, we cannot determine unequivocally whether scoliosis leads to higher shoulder asymmetry or whether higher shoulder asymmetry leads to scoliosis. If scoliosis
were the only factor leading to higher shoulder asymmetry, then the direction of the spinal curve should correspond significantly to the direction of higher shoulder asymmetry. Because it did not, we conclude that the positional asymmetry may, in part, be causing scoliosis. We propose that a combination of orthopedic asymmetries, unique for each person, results in the wide variety of spinal curve patterns and positional asymmetries seen in Rett syndrome (see Figure 1).

The finding that total asymmetry affected the prevalence of scoliosis in Rett syndrome suggests an important area for further investigation. Longitudinal studies to periodically evaluate both curve progression and asymmetry of movement and position should help to clarify this relationship.

Out logistic regression analysis indicates that ambulation protects against Rett syndrome scoliosis and, furthermore, that lack of ambulation was an important factor in predicting scoliosis. Symmetric movements and positioning were protective against scoliosis in participants who were ambulatory (see Table 3). In participants who were nonambulatory, symmetric positioning but not symmetric movements were protective against scoliosis. Symmetric bodyside movements may be protective, but a larger sample size is needed to confirm this. The activity of ambulation with reciprocal weight shift and alternating arm swing should contribute to a bilaterally symmetric muscle pull, protecting against scoliosis. Maintaining ambulation also allows more independence for the child and less physical effort from family members to care for the child. Our finding that ambulation is associated with a lower incidence of scoliosis suggests that maintenance of functional ambulatory skills throughout daily living tasks should be a primary area of occupational therapy intervention.

The prevalence of Rett syndrome scoliosis increased dramatically with age (see Table 1), replicating Basset and Tolo’s (1990) finding, but our use of logistic regression analysis provides more robust support for this finding. The increase of the odds ratio with the older age groups is another indication that the older the child, the more likely the child will develop scoliosis. We believe that this association of scoliosis with advancement of age may be due to the continual, asymmetric muscular pull on the spine over time. That the severity of the curve increased in the older age group indicates a progression of scoliosis with age. This finding suggests that occupational therapy practitioners be aggressive in their efforts at an early stage in the disease process and educate family members in the proper strategies, for example, to be used with functional tasks in the home.

Evidence of the hemisyndrome aspect of Rett syndrome is sketchy in the literature. We await further studies to supplement our finding of prevalence for right scoliosis and right asymmetry and Budden’s (1995) finding of right lower-extremity weakness.

Intensive therapeutic intervention of CNS symptoms, including scoliosis, are appropriately based on the hypothesis of neurodevelopmental arrest etiology of Rett syndrome (Armstrong, 1995) and are reported to be successful (Budden, 1995; Glaze, 1995; Hanks, 1990). Therapy goals for neurodevelopmental disorders can be developed with the expectation of some improvement in level of functioning, whereas therapy goals for neurodegenerative disorders would focus on maintaining current levels of functioning. Hanks (1986, 1990) described a detailed treatment program for Rett syndrome scoliosis, including developing righting and equilibrium reactions, improving ambulation, and maintaining mobility. Budden (1995) recommended treatment based on decreasing repetitive stereotypic movements (e.g., by using splints) as well as intensive therapy for the scoliosis, apraxia, and rigidity. She reported that none of the patients in her study required surgery for scoliosis but were seen clinically for therapy and were monitored closely for progression of scoliosis. Lieb-Lundell (1988) recommended treatment programs be based on the stage of progression of Rett syndrome.

Upper-extremity, sensory input treatments that are based on the neurophysiological principles of Rood (as cited in Huss, 1983), including hand splinting (Kubas, 1992; Naganuma & Billingsley, 1983) and elbow orthoses (Aron, 1990; Sharpe, 1992), have been reported to decrease the frequency and occurrence of the abnormal, stereotypic, nonvolitional hand and arm movements. Coincidentally, these treatments increased ambulatory skills and ability to engage in fine and gross motor functional tasks if one hand was held or limited in movement in order to free the child with Rett syndrome from the nonvolitional, stereotypic movement focus (Hanks, 1986; Witt-Engerstrom, 1992). If these hand splints and elbow orthoses can help encourage functional upper-extremity motor skills and ambulation, they should help to decrease the severity and progression of scoliosis.

Limitations

There are several limitations to this study. First, it evaluated orthopedic risk factors at the one fixed time when the survey was completed and not over time. Longitudinal studies of closely monitored girls with Rett syndrome could give more detailed symptom progression information. Second, the study relied on information provided by nonprofessional caregivers about abnormal movements, ambulation, and scoliosis of the child with Rett syndrome. However, respondents used the “I don’t know”
category for each question infrequently (.6% of the responses), indicating a high confidence among caregivers regarding their ability to complete the questionnaire accurately. The prevalence of Rett syndrome scoliosis found in this study was similar to that obtained by previous studies that were based on physician reports and radiographs (Bassett & Tolo, 1990; Harrison & Webb, 1990), indicating that the caregivers provided accurate data regarding the prevalence of scoliosis. Finally, some respondents reported symmetry that may not have included the asymmetric, hand dominance aspect of that child.

Conclusion
In Rett syndrome, asymmetries in movement and position correlated with scoliosis; symmetry in movement and position correlated with no scoliosis. Age; asymmetric, abnormal upper-body positioning; and inability to ambulate were significant predictors of scoliosis. Asymmetric body side and combined movements were supporting factors of scoliosis. Right total asymmetry and higher shoulder asymmetry showed an expected higher prevalence of right direction of the scoliosis curve, and, correspondingly, left asymmetries were more prevalent with left scoliosis.

This study has given the first rationale for a noninvasive treatment of Rett syndrome scoliosis that focuses on balancing the bilateral muscular pull and overriding the stereotypy focus. A longitudinal study is recommended to evaluate scoliosis along with body movements and positions and orthopedic and sensory motor treatments.

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References


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