Rehabilitation Management in patients with Rett Syndrome

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Disclosures
The authors have nothing to disclose

Objectives
Enumerate the common impairments with Rett Syndrome and rehabilitation strategies
Discuss principles and options for dystonia management
Rett Syndrome (RTT)

- A neurodevelopmental disorder
- 1:10,000 predominantly affecting females
- Apparent normal initial development followed by regression of fine motor and communication skills (6-18 months of age)
- 80-90% of Classic RTT has mutation of MECP2 (methyl-CpG-binding protein 2) gene, at the tip of x chromosome.
  > 200 mutations identified (varied size of deletion)
- Not a neurodegenerative, nor a progressive disease
- A clinical diagnosis

Variant or Atypical RTT

- May have some clinical features, not all

  CLINICAL SEVERITY score
  QOL with clinical severity
Clinical/Disease profile: distinct pattern of disability, consistent regardless of level

- Impaired hand use and speech
- Disturbed posture and balance
- Defective central processing of signals
- Generally normal peripheral reception of sight and sound
- Disturbed heart rate, blood pressure and respiratory rhythm

Used to describe: Four Stages of RTT

**Stage I (Early Onset)**
- 6 and 18 months
- Symptoms vague
- Less eye contact
- Gross motor skills delayed
- Hand wringing

**Stage II (Rapid Destructive)**
- 1 and 4 years
- Gradual or rapid onset
- Loss of purposeful hand skills and spoken language
- Gross motor skills delayed
- Trouble initiating movements

**Stage III (Plateau)**
- 2 and 10 years
- Apraxia
- Continued motor problems
- Seizures
- Improved behavior

**Stage IV (Late Motor Deterioration)**
- Reduced mobility
- Muscle weakness
- Scoliosis
- Hypertonia/Rigidity/Spasticity
- May stop walking

Table 1: Aspirated developmental milestones: gross and fine motor

<table>
<thead>
<tr>
<th>SIT</th>
<th>Absent (0-5yr old)</th>
<th>Absent (6-10yr old)</th>
<th>Absent (11-15yr old)</th>
<th>Absent (16-20yr old)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Absent</td>
<td>Tactile (0-5yr old)</td>
<td>Tactile (6-10yr old)</td>
<td>Tactile (11-15yr old)</td>
<td>Tactile (16-20yr old)</td>
</tr>
<tr>
<td>Tactile</td>
<td>Visual (0-5yr old)</td>
<td>Visual (6-10yr old)</td>
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<td>Visual (16-20yr old)</td>
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<tr>
<td>Visual</td>
<td>Auditory (0-5yr old)</td>
<td>Auditory (6-10yr old)</td>
<td>Auditory (11-15yr old)</td>
<td>Auditory (16-20yr old)</td>
</tr>
<tr>
<td>Auditory</td>
<td>Motor (0-5yr old)</td>
<td>Motor (6-10yr old)</td>
<td>Motor (11-15yr old)</td>
<td>Motor (16-20yr old)</td>
</tr>
<tr>
<td>Motor</td>
<td>Fine (0-5yr old)</td>
<td>Fine (6-10yr old)</td>
<td>Fine (11-15yr old)</td>
<td>Fine (16-20yr old)</td>
</tr>
<tr>
<td>Fine</td>
<td>Gross (0-5yr old)</td>
<td>Gross (6-10yr old)</td>
<td>Gross (11-15yr old)</td>
<td>Gross (16-20yr old)</td>
</tr>
<tr>
<td>Gross</td>
<td>All (0-5yr old)</td>
<td>All (6-10yr old)</td>
<td>All (11-15yr old)</td>
<td>All (16-20yr old)</td>
</tr>
</tbody>
</table>

Common clinical findings and impairments

- Postural instability/proximal hypotonia
- Distal hypertonia/dystonia (may generalize w/ age)
- Apraxia
- Altered sensation/ impaired proprioception, Ataxic gait
- Limited dexterity/fine motor skills
- Stereotypic movements
- Seizures (60%)
- Muscle Weakness

Common clinical findings and impairments

- Head growth deceleration, Growth retardation
- Muscle imbalances
- Scoliosis/kyphosis (>50%)
- Joint contractures
- Impaired sleep pattern
- Oral/speech problems
- Respiratory problems
- GI & Nutrition problems (GI dysmotility, feeding and swallowing problems)
- Personality is well-preserved, relates warmly with other people, music helps

Multi- and Inter- disciplinary Team

- Blue Bird Rett Center (Neurology)
- Orthopedics
- GI, Nutrition
- PM&R and Rehab team
  - Physical Therapist
  - Occupational Therapist
  - Speech and Language Therapist

... and of course, the family
Functional performance may be related to type of genetic defect

Importance of assessing specific need of each patient in structuring early intervention rehab program

Understanding relationship between mutations and functional skills may enhance ability to assist, counsel families in developing appropriate rehab goals and strategies

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**PT Plan of Care**

- Goals should be individualized
- ROM
  - Daily!
  - Family participation
  - Repetition is key!
- Postural stability
- Strengthening:
  - Proximal—Distal
  - Static Positioning
  - Therapeutic Exercise

**OT Plan of Care**

- Focus is usually upper limbs and daily activities
- Strengthening
- Coordination and postural control
- Dyspraxia
- Access to augmentative communication
- Adaptive aids
- Oromotor skills

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**Speech Therapy**

- Oromotor skills
- Improve chewing and swallowing
- Improve handling of secretions
- Bruxism
- Communication
- Receptive and expressive language
Communication

Not being able to talk is not the same as not having anything to say

Rosemary Crossley

Ask for therapist with experience in aug comm

School can bring in AT consultant

Family Participation

Practice, practice, practice

Repetition is key

Therapist provides feedback about technique/positioning, alternate strategies, willing to change directions/approaches

Family/caregiver involvement on daily basis is a must be done daily

Alternative Therapies

Hippotherapy

Aquatherapy

Music Therapy

Art

Massage
Orthoses

- Support/stabilize joints
  - Ankle foot orthoses, supramalleolar orthosis
  - Hip alignment and trunk control (compression garments)
- Prevent/limit deformity – large joints may require stiffer plastic and inner boot

Orthoses, cont’d

Rehab Equipments

Supportive seating
Gait/mobility aids
Stander
Lift system
Identify goals for tone management

- Prevent worsening/progressing of contractures and deformity
- Improve positioning/posture
- Ease of care (dressing, diapering)
- Improve bracing tolerance
- Prevent skin breakdown
- Impede mobility, transfers and function

Traditional Step-Wise Approach to Treat Hypertonia

- orthopedic procedures
- Chemoneurolysis (botulinum toxin/phenol)
- Oral medications for hypertonia
- Rehab Therapies/Orthoses
- Assess for medical reason for change in tone and/or remove

Oral medications for hypertonia

- Take into consideration potential side effects
  - Sedation
  - Decreased postural control
  - Increased drooling
  - Constipation and GI upset
  - Interaction with other medications
- Some girls are very sensitive while others have minimal to no side effects
- In severe cases, usually meds help some but not enough and maximum doses are reached due to side effects
  - Common meds used: baclofen, diazepam, clonazepam, tizanidine, trihexyphenidyl (case report for ALTE due to dystonia)
**Chemoneurolysis**

Botulinum toxin injection
- may be repeated every 3-6 months

Phenol 5-6% to motor branch (Obturator, musculocutaneous, tibial nerves)

**Intrathecal baclofen pump**

For global/generalized tone reduction in patients with severe spasticity or dystonia
Catheter placement will impact degree of tone reduction
Implanted system directs concentrated baclofen to the CSF
TCH ITB program:
3 RTT (T158M, 806 delG, Ex 3&4 del) patients with ITB pumps, 18-29 y/o, dose 100-1000mcg/day

**Goals Should be Individualized**

- Every child is different...
- Some girls have greater deficits in strength or tonal abnormalities than others
- Some have medical issues that will impact therapy participation
  - Anxiety, seizures, pain
- One therapy approach may work for one child and not for another – be flexible
- Therapists should be willing to seek out suggestions and collaborate with each other (same facility or other)
- While there may be similarities in approaches to children with CP, Rett girls are different!
References:


Special Thanks

To our patients and families who graciously consented to share photographs and videos

Assessment and case study of a patient with Rett Syndrome

Carla Uria PT
Objectives

- Recognize common motor impairments in children with Rett syndrome and how they affect their ability to function on a daily basis.

- Establish appropriate functional goals that apply to their specific and current limitations

Specific Motor Problems

- Postural instability
- Hypotonia
- Ataxia
- Apraxia/Dyspraxia
- Loss of transitional movements
- Impaired gait /loss of gait or never walk
- Loss of hand function
- Dystonia
- Foot deformities equino varus
- Kyphosis/Scoliosis
- Distal Hypertonia

Impairments

- Poor registration discrimination to tactile and proprioceptive information
- Visual and proprioceptive deficits = Spatial disorientation
- Decreased sensory modulation = fear to move
- Decreased kinesthetic memory
Other impairments affecting motor function

- Respiratory problems = holding breath patterns/decrease ribcage mobility.
- Cardiovascular changes = cool or warm feet or hands with change of color
- Decreased communication (i.e. eye contact)
- Irritability and anxiety

Physical Therapy Goals

- Maintain or increase motor skills
- Develop or maintain transitional skills
- Prevent or reduce deformities
- Alleviate discomfort and irritability
- Improve independence
- Develop Home Exercise Program/Assure family comfortable with exercises

Goal Setting

Considerations

- Patient - functional abilities - impairments
- Family expectations
- Set Individualize/unique goals according current functional status
- Two methods of goals setting: SMART and GAS( goal attainment scale)
SMART Goals

- **Specific/Significant:** Who-recipient/family/caregiver
- **Measurable/meaningful:** (i.e., with metrics/criteria of mastery? How well?)
- **Achievable/Action Oriented:** What functional behavior/skill? Be realistic
- **Realistic/Relevant:** Context/setting Specific conditions
- **Timely/Trackable:** How often?

Goal Attainment Scale (GAS)

- GAS is a useful tool for measuring achievement of goals once the areas for goal setting are established. (Kiresuk & Sherman, 1968, Kiresuk 1994; King 1999)
- Goals should be relevant, understandable, measurable, behavioral, attainable, and time-related.
- The GAS procedure involves:
  - **unique goal**
  - **five level scale**
  - **evaluate**
- A key element: only one component of the behavior can be measured per goal at a time.

GAS Goals

<table>
<thead>
<tr>
<th>Grade/Level</th>
<th>Definition</th>
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</thead>
<tbody>
<tr>
<td>-2</td>
<td>Much less than expected outcome (Baseline)</td>
</tr>
<tr>
<td>-1</td>
<td>Somewhat less than expected outcome</td>
</tr>
<tr>
<td>0</td>
<td>Expected outcome</td>
</tr>
<tr>
<td>+1</td>
<td>Somewhat more than expected outcome</td>
</tr>
<tr>
<td>+2</td>
<td>Much more than expected outcome</td>
</tr>
</tbody>
</table>
Goal setting

SMART Goal

The patient will walk 15 feet from the bedroom to the bathroom with one hand held assistance and verbal cues every morning to brush her teeth 5/7 days per week.

<table>
<thead>
<tr>
<th>Level</th>
<th>GAS Goal</th>
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</thead>
<tbody>
<tr>
<td>-2</td>
<td>Walks 3 feet</td>
</tr>
<tr>
<td>-1</td>
<td>Walks 10 feet</td>
</tr>
<tr>
<td>0</td>
<td>Will walk 15 feet</td>
</tr>
<tr>
<td>+1</td>
<td>Walks 20 feet</td>
</tr>
<tr>
<td>+2</td>
<td>Walks 25 feet</td>
</tr>
</tbody>
</table>

Case Study History

- Name: Noemi
- Age: 3 years old
- Diagnosis: Rett Syndrome
- PT evaluation at TCH on May 2013
- Born full term uncomplicated pregnancy.
- Gross motor deficits
- Crawled briefly backwards at 8 months
- Did not achieve sitting pull to stand
- Grasped and held on to objects.
- Babble, smile and interact with parents up to 8 months
- Steady decline with communication and interaction and lost by 20 months
Case Study History (continued)

- Developed repetitive finger rubbing and later hand mouthing with right hand movements.
- Became social and interactive again at 30 months of age.
- Received physical therapy, mainly ROM and Feldenkrais technique.
- Orthotics full body compression garment (DMO) and Posterior Leaf AFO
- Lives in Italy with parents

History

- Family arrived to Houston seeking multidisciplinary evaluation with Rett syndrome team and HEP
- Parent concerns:
  - Irritability
  - Poor tolerance to exercises and orthotics
  - No initiation of movement.

Observation/Function

- Happy, smiles making eye contact present
- Appears to understand some commands
- Enjoys music
- Right hand on her mouth often
- Frequent hand movements
- Sits with support, but no initiation of transitions
- Anxious when therapist attempted weight shifts with teeth grinding
- Family not comfortable assisting with mobility and HEP
Posture

- Decreased postural/trunk control, decreased mobility
- Head forward
- Wide base of support
- Inefficient balance with increased activation of distal musculature.

Muscle tone

- Hypotonia - trunk and proximal joint musculature
- Increase LE extensor tone (gastrocs – soleus, quadriceps)
- Dystonia hands, feet, tongue

Musculoskeletal

- **Muscle Strength**: Trunk, UES and LES weakness, moves all 4 limbs spontaneously
- **ROM**: Full PROM. No purposeful UE movement
- No scoliosis/spinal deformities
Gross motor activities

- **Rolling** - max A for LEs, trunk support and UE positioning.
- **Ring sitting** - maintains for 2-3 seconds before falling forward when supported at hips.
- **Short sitting** - in cube chair with CGA/MinA for up to 20 seconds
- **Stands** - with mod A at hips for 6 seconds before trunk falls forward flexed at hips.
- Significantly impaired gross motor development per HELP Chart (2 to 3 months)

Transfers

- Transfers with max assistance
- Sit to stand from physioball with max A at hips

Sensory

- Decreased tactile and proprioception at trunk UES, LES proximally
- Decreased sensory modulation
- Decreased Kinesthesia
GAIT

• 10 steps with max A for standing with facilitation at hips + tactile to maintain trunk extension to prevent falling forward with whole body compression garment donned.

• Required max A for stepping and was unable to initiate steps with head extended, UEs abducted at the shoulder with elbows flexed, hands fisted.

GAIT

• Wide base of support
• Short steps
• Ataxia
• Sway movements of trunk
• Tendency to fall backwards
• Problem initiating steps

Short Term Goals SMART

Initial
Patient will maintain ring sitting with upright trunk for up to 15 seconds with assistance at hips or lower 3/5 trials for safe and I sitting.

Progression at one month
Patient will maintain ring sitting on mat with upright trunk for up to 5 seconds with assistance at hips or lower 3/5 trials for safe and I sitting.
Short Term Goals SMART

**Initial**

Patient will maintain side sitting on firm surface with arm propping x 30 seconds with CGA 2/4 trials in preparation to initiate transitions.

**Progression at one month**

Pt will maintain side sitting on firm surface with arm propping x 30 sec with SBA 2/4 trials in preparation to initiate transitions.

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Short Term Goals SMART

**Initial**

Patient will static stand 10 seconds with compression garment (DMO)and AFOS donned with facilitation at hips 3/5 attempts for increased I in transfers.

**Progression at one month**

Patient will static stand 20 seconds with compression garment (DMO)and AFOS donned with facilitation at knees 3/5 attempts for increased I in transfers.

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Short Term Goals SMART

**Initial**

Guided by therapist the caregiver will be able to perform ROM, assist with transitions with minimal tactile cues for hand position and appropriate amount of sensory input to progress towards I with HEP.

**Progression at one month**

Caregiver will assist Noemi with (ROM, transitions, sensory input) to progress toward I with HEP with verbal cueing from the therapist.
<table>
<thead>
<tr>
<th>Grade</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>-2 baseline</td>
<td>Patient maintains ring sitting for 2-3 seconds before falling forward when supported at hips</td>
</tr>
<tr>
<td>-1 progress</td>
<td>Patient will maintain ring sitting with upright trunk for up to 5-10 seconds with assistance at hips or lower 3/5 attempts for safe and I sitting.</td>
</tr>
<tr>
<td>0 Goal</td>
<td>Patient will maintain ring sitting with upright trunk for up to 15 seconds with assistance at hips or lower 3/5 attempts for safe and I sitting.</td>
</tr>
<tr>
<td>+1 somewhat more than expected</td>
<td>Patient will maintain ring sitting with upright trunk for up to 16-25 seconds with assistance at hips or lower 3/5 attempts for safe and I sitting.</td>
</tr>
<tr>
<td>+2 more than expected</td>
<td>Patient will maintain ring sitting with upright trunk for more than 26 seconds with assistance at hips or lower 3/5 attempts for safe and I sitting.</td>
</tr>
</tbody>
</table>

**Long Term Goals (SMART)**

1. Patient will tolerate standing on BOSU ball 30 seconds x 3 with min A facilitation at trunk with AFO donned to improve dynamic standing balance and reduce anxiety during weight shifting.
2. Patient will be able to ambulate x 20 steps with mod A, facilitation at trunk, with patient initiating 50% of stepping for walking short distances in home.
3. Patient will maintain side sitting with UE propping x1 minute when placed, with SBA when stimulated by toy 2/4 trials to encourage play.
4. Caregiver will report compliance with current HEP 5 days a week.

**6 months later**

- Patient met all goals
- Family/patient returned to Italy
- Mother was comfortable with HEP, facilitating transitions and static standing and ambulation.
- Patient was wearing compression garment and solid AFOS 8 hours a day and short leg cast splints during the night.
- Noemi enjoyed vestibular input (swing, tricycle)
After 10 months

Noemi returned to Houston having maintained her previous level:

- Sitting - side sit for 20 sec at a time.
- Standing - static standing with CGA at hips for several minutes,
- Gait-ambulated 15 to 20 ft. at a time with min to mod assist at hips initiating the steps consistently.
- Tone- LE extensor tone in gastrosoleus, quadriceps, and in gluteals to a lesser degree.
- Gross motor skills- level of a 6 to 8 month old child assessed on HELP scale used for measurement of gross motor development.

References

Augmentative and Alternative Communication (AAC) and Rett Syndrome

Betsy Furler, MS, CCC-SLP
What is Communication?

- Communication is simply the act of transferring information from one place to another.
  
  [http://www.skillsyouneed.co.uk/IPS/What_is_Communication.html#ixzz29f4fSRl](http://www.skillsyouneed.co.uk/IPS/What_is_Communication.html#ixzz29f4fSRl)

- There are many types of communication.
  
  - Verbal
  
  - Nonverbal - gestures, body language, facial expressions, dress
  
  - Written
What is AAC?

- Devices available
- Apps available
- Tools and strategies that help people communicate
- Reasons for difficulty with communication: motor impairments, language difficulties, voice disorders
- Goals: Independence, Self Advocacy as well as Functional Communication
First Steps

- Locate a qualified professional for the assessment
- Define goals and expectations
- Look at needed communication and settings where the user needs to communicate
- Determine appropriate strategy and/or device
- Program and implement the strategy and/or device
• Full Featured Symbol Based Devices

• Promotes syntax and vocabulary development, the symbol system includes words from all parts of speech and the ability to use and link multiple pages.

• A variety of high quality voices

• An available keyboard to increase the variety of communication.
**Types of AAC Devices**

- Mid Featured AAC Devices
- Includes a symbol set with verbs, adjectives and nouns.
- Pre-programmed pages for basic wants/needs.
Types of AAC Devices

- Basic communication apps
- Allows limited basic communication
- Includes primarily nouns with some rote phrases such as “I want...”
- Most utilize recorded voices
Types of AAC Devices/Strategies

• Simple Communication - Yes/No - 2-12 choices on one page
• Scene based communication
• Text based communication
Myths and Fears About AAC

http://aac.unl.edu/yaack/b2.html - YAACK - AAC Connecting Young Kids

• Myth - AAC should be introduced only after giving up hope of natural speech

• Truth - AAC should be used with speech therapy in order to encourage the child to use any means of communication available

• Beukelman & Mirenda, 1992
Myths and Fears About AAC

- Myth - The use of AAC reduces the child’s desire to use speech to communicate.

- Truth - As children learn to communicate with AAC, they are more motivated to communicate through all means, including speech.

- Beukelman & Mirenda, 1992; Silverman, 1980; Van Tatenhove, 1987
Myths and Fears About AAC

http://aac.unl.edu/yaack/b2.html - YAACK - AAC Connecting Young Kids

• Myth - A young child is not ready for AAC.

• Truth - Typically developing children use a variety of methods of communication starting shortly after birth. AAC can be used starting at a very young age when the system is tailored to the developmental level of the child.

• Reichle, York, & Sigafoos, 1991
Why Use the iPad for AAC?

• Pros

• Low cost and easily acquired

• The “cool” factor

• Can be used for temporary disabilities

• Multi-functional - one device can be used for a variety of AAC functions as well as organizational and academic needs

• Easy to change apps - lower cost than changing a device
Why Use the iPad for AAC?

• Pros

• Easy repairs in many big cities - screen replacement as low as $113

• Replacement less expensive - $500 for the iPad (less if insured) and apps can be reloaded at no additional cost

• Availability of substitute devices - iPod Touch or iPhone

• Highly portable - lightweight even for a young child.
Why Use the iPad for AAC?

- The device is very interactive, responds quickly and is engaging.
- Everything is more fun on an iPad!
- Key guards are available for most AAC apps - http://www.laseredpics.biz/servlet/the-Adaptive-Technologies-Supplies/Categories
Why Not To Use the iPad for AAC?

• Cons

• Customer/Technical Support may be limited

• Not dedicated devices for individuals

• Limited training and support

• Lack of insurance coverage

• Access issues for some users - no eye gaze technology

• Limited switch access
Where To Start

- Can the user access the device?
- What size of array is the user successful with?
- AAC Evaluation Genie - evaluation app for AAC use
- What is cognitive level? Can the user understand line drawings? Do they need photos?
- Can the user read? Type?
- In what environments will the device be used in?
Remember Cognitive Concerns

- Match device/app and expectations to cognitive level
- Start at the “just right level” but allow for growth
- Don’t underestimate the child’s abilities! This is very important with girls with Rett Syndrome.
- Use age appropriate types of communication
- Children should begin with play on the AAC system because typical children use language primarily for play.
Photos, Symbols, Text

• Should you use photos, symbols or text?

• Real life color photos of the child’s environment are the easiest to understand.

• Next - generic color photos

• Black and white photos add a degree of difficulty.

• Then color drawings

• Black and white line drawings

• Symbol based systems.

• Text - must be literate to use text successfully.
Simplifying An Advanced Page or User Set

- Decrease the number of buttons on each page.
- Consider using photos rather than drawings for the icons.
- Decrease the number of screens needed for each message.
Introducing A New Device

• Allow time for exploration - http://adaptingcreatively.blogspot.com/2011/02/case-for-exploration-aac-style.html?m=1

• Use games/play to orient child to the new app

• Make it fun - a child’s work is play - they should be encouraged to play and communicate during the play.
AAC Must Be A Part of Life

- Success is built on repetition.
- The child must have access to the device at all times of the day and in all settings.
- The child must be allowed to “play with” or explore the device.
- The adults in the child’s life must buy into the importance of the device and model the use of the device/app.
Resources for AAC Apps

- www.bridgingapps.org
- List of apps: http://bridgingapps.org/list/?id=45793
- Insignio - search engine
- We have reviewed 63 AAC apps and have over 100 listed.
- New apps are coming out all the time!
Resources for Funding

- STAP Program - [http://www.dars.state.tx.us/dhhs/stap.shtml](http://www.dars.state.tx.us/dhhs/stap.shtml)
- Insurance
- Medicaid
- Silent Stars - [www.silentstars.org](http://www.silentstars.org) - AAC for Apraxia
Questions?

- Betsy Furler, MS, CCC-SLP
- [www.bridgingapps.org](http://www.bridgingapps.org)
- bfurler@sbcglobal.net
- Twitter: BetsyFurler and BridgingApps
- Facebook: Betsy Walling Furler