

An exploration of the use of eye gaze & gestures in females with Rett syndrome

Anna Urbanowicz

a.urbanowicz@uq.edu.au



Proudly supported by the people of Western Australia through Channel 7's Telethon

Background



Communication

- Critical to participation in everyday life
- Challenging for people with severe disabilities
 - Non-verbal communication modalities:
 - Vocalisations
 - Body movements
 - Eye gaze
 - Gestures

– Communication partner interpretation

(Duker, van Driel, & van de Bercken, 2002; Maes, Vos, & Penne, 2010)





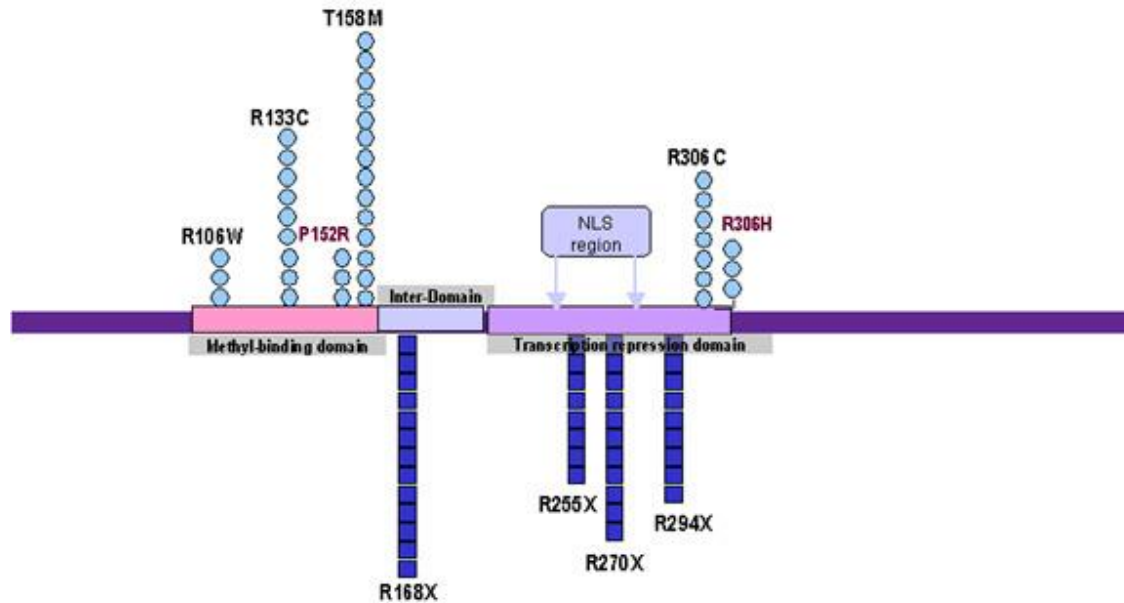
Diagnostic criteria for Rett syndrome

Main criteria	Supporting criteria
<ul style="list-style-type: none">• A period of regression:<ul style="list-style-type: none">• Loss of purposeful hand skills• Loss of spoken language• Gait abnormalities• Stereotypic hand movements	<ul style="list-style-type: none">• Breathing disturbances• Bruxism• Sleep problems• Abnormal muscle tone• Scoliosis/kyphosis• Growth restriction• Small cold hands & feet• Laughing/screaming spells• Diminished response to pain• Intense eye communication

(Neul et al., 2010)



Cause of Rett syndrome



- *MECP2* mutations have been linked to the cause of typical Rett syndrome in the majority of cases (Amir et al., 1999)

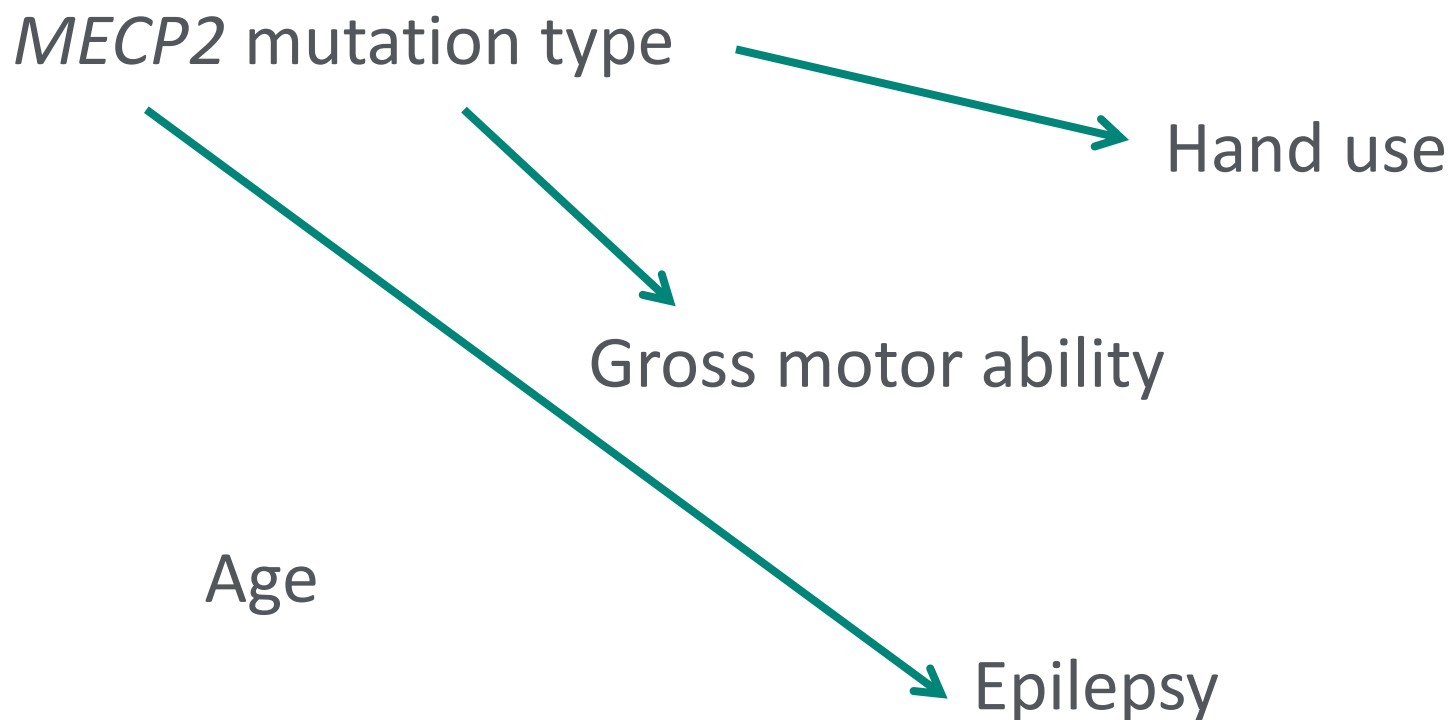


Eye gaze & gestures in Rett syndrome

- Few females with Rett syndrome use words (Urbanowicz et al., 2014)
- Historically eye gaze has been a communicative strength (Hagberg, 1995)
- Gestures are used less than eye gaze (Bartolotta et al., 2011)
- *But why do only a small proportion use gestures in comparison to eye gaze?*



Factors influencing eye gaze & gestures



(Bebbington et al., 2008, Didden, et al., 2010)



What we wanted to know

- How often do girls and women with Rett syndrome use eye gaze and gestures for communication?
- Do genotype, age and motor abilities influence the use of eye gaze and gestures?



Methods



Australian Rett Syndrome Database

Established 1993



Initial Family & Clinician Questionnaires

- Pregnancy & child's birth, early development & current level of functioning



Follow-up Family Questionnaires

- Completed in 2000, 2002, 2004, 2006, 2009 & 2012
- Medical conditions & care, everyday functioning, specific Rett syndrome behaviours, use of resources & family functioning



Video-based Evaluation & Parent-report checklist

- Completed in 2004/2005 & 2007/2008
- Communication, eating & drinking, hand movements & functions, personal care & Rett syndrome specific behaviours





Communication outcome

- Parent-report checklist
- Communication and Symbolic Behavior Scales
Developmental Profile: Infant-Toddler Checklist
(Wetherby & Prizant, 2002)
 - Eye cluster score (best score = 6)
 - Gestures cluster score (best score = 10)





Independent variables

- *MECP2* mutation type
- Age group:
 - < 8 years
 - 8 < 13 years
 - 13 < 19 years
 - > 19 years
- Gross motor abilities:
 - General gross motor abilities
 - Complex gross motor abilities



Data analysis

- Multivariate linear regression
 - Relationships between eye gaze and gestures scores & independent variables



Results



Sample characteristics

- Age range 2.3 – 33.68 years
- Mean total gross motor score was $32.17 \pm 14.81/60$
 - Mean general gross motor score $24.20 \pm 11.31/40$
 - Mean complex gross motor score $7.97 \pm 4.22/20$





CSBS DP ITC items

Item	Frequency of behaviour % (n)		
	Often	Sometimes	Not yet
Emotion and Eye Gaze			
Do you know when your daughter is happy and when she is upset?	93.92% (139/148)	6.08% (9/148)	0% (0/148)
When your daughter plays with toys (or some other objects), does she look at you to see if you are watching?	30.00% (42/140)	49.29% (69/140)	20.71% (29/140)
Does your daughter smile or laugh when looking at you?	72.97% (108/148)	25.00% (37/148)	2.03% (3/148)
When you look at and point to a toy (or some other object) across the room, does she look at it?	20.27% (30/148)	60.81% (90/148)	18.92% (28/148)
Communication			
Does your daughter let you know she needs help or wants an object out of reach?	30.00% (45/150)	46.67% (70/150)	23.33% (35/150)
When you are not paying attention to your daughter, does she try to get your attention?	44.37% (67/151)	45.70% (69/151)	9.93% (15/151)
Gestures			
Does your daughter pick up objects and give them to you?	9.46% (14/148)	12.84% (19/148)	77.70% (115/148)
Does your daughter show objects to you without giving you the object?	6.8% (10/147)	17.01% (25/147)	76.19% (112/147)
Does your daughter wave to greet people?	1.37% (2/146)	9.59% (14/146)	89.04% (130/146)
Does your daughter point to objects?	10.20% (15/147)	29.93% (44/147)	59.86% (88/147)
Does your daughter nod her head to indicate yes?	4.76% (7/147)	13.60% (20/147)	81.63% (120/147)
Sounds			
Does your daughter use sounds or words to get your attention or help?	40.14% (59/147)	46.26% (68/147)	13.60% (20/147)
Does your daughter string sounds together, such as uh oh, mama, or bye bye?	9.59% (14/146)	26.03% (38/146)	54.38% (94/146)
Understanding			
When you call your daughter's name, does she respond by looking or turning to you?	60.14% (89/148)	38.51% (57/148)	1.35% (2/148)



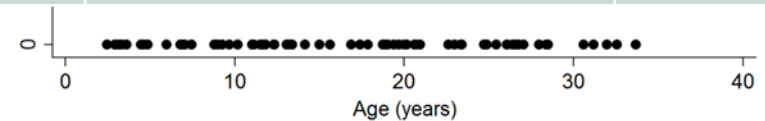
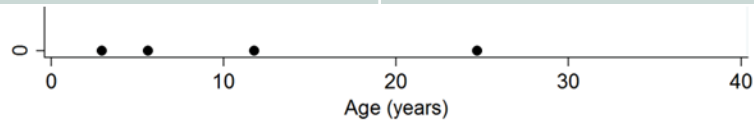


Eye gaze & gestures scores according to age (n=151)

Eye gaze

Gestures

Age group (n)	Eye gaze		Gestures	
	Coefficient (95% CI)	p	Coefficient (95% CI)	p
< 8 years (32)	0.22 (-0.38 – 0.83)	0.47	0.31 (-0.63 – 1.22)	0.50
8 < 13 years (37)	0.72 (0.14 – 1.30)	0.02	0.50 (-0.33 – 1.33)	0.23
13 < 19 years (29)	0.87 (0.24 – 1.50)	0.01	0.45 (-0.48 – 1.38)	0.34
≥ 19 years (53)	Baseline	-	Baseline	-



Eye gaze & gestures scores according to genotype (n=151)

Mutation type (n)	Eye gaze		Gestures	
	Coefficient (95% CI)	p	Coefficient (95% CI)	p
p.ArgR168* (16)	Baseline	-	Baseline	-
large deletion (11)	-0.38 (-1.48 - 0.71)	0.49	0.14 (-1.36 -1.65)	0.85
p.ArgR106Trp (9)	-0.24 (-1.41 - 0.93)	0.68	-0.66 (-2.27 -0.94)	0.42
p.Arg133Cys (12)	0.23 (-0.84 - 1.30)	0.67	1.38 (-0.10 -2.86)	0.07
p.Arg255* (13)	0.39 (-0.66 - 1.44)	0.46	0.03 (-1.41 -1.48)	0.97
p.Arg270* (10)	0.51 (-0.62 - 1.64)	0.37	0.51 (-1.04 -2.05)	0.52
p.Arg294* (12)	0.48 (-0.59 - 1.64)	0.37	0.69 (-0.78 -2.17)	0.36
p.Arg306Cys (10)	-0.05 (-0.12 - 1.08)	0.92	0.25 (-1.30 -1.81)	0.75
p.Thr158Met (14)	-0.47 (-1.50 - 0.55)	0.36	-0.26 (-1.66 -1.14)	0.71
C-terminal deletion (16)	0.29 (-0.70 - 1.28)	0.58	1.53 (0.17 -2.89)	0.03
Early truncation (7)	0.17 (-1.10 - 1.54)	0.79	-0.64 (-2.38 -1.09)	0.47
Other (21)	-0.21 (-1.14 - 0.72)	0.65	0.08 (-1.21 - 1.36)	0.91

Eye gaze & gestures according to motor abilities (n=117)

	Eye gaze		Gestures	
	Coefficient (95% CI)	p	Coefficient (95% CI)	p
General gross motor (n)				
Average & below (58)	Baseline	-	Baseline	-
Above average (59)	0.40 (-0.20 – 1.00)	0.188	1.70 (0.94 – 2.47)	<0.001
Complex gross motor (n)				
Average & below (74)	Baseline	-	Baseline	-
Above average (43)	0.68 (0.06 – 1.30)	0.033	2.25 (1.49 – 3.00)	<0.001

Conclusions



Summary

- Eye gaze > gestures
- Older women used eye gaze less
- Females with better than average motor abilities used gestures more
- On average, eye gaze was used frequently across mutation groups
- On average, gestures were used more frequently by females with p.Arg133Cys mutations or C-terminal deletions





References

- Amir, R. E., Van den Veyver, I. B., Wan, M., Tran, C. Q., Francke, U., & Zoghbi, H. Y. (1999). Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2. *Nature Genetics*, 23(2), 185-188. doi: 10.1038/13810
- Bartolotta, T. E., Zipp, G. P., Simpkins, S. D., & Glazewski, B. (2011). Communication skills in girls with Rett syndrome. *Focus on Autism and Other Developmental Disabilities*, 26(1), 15-24. doi: 10.1177/1088357610380042
- Bebbington, A., Anderson, A., Ravine, D., Fyfe, S., Pineda, M., de Klerk, N., . . . Leonard, H. (2008). Investigating genotype-phenotype relationships in Rett syndrome using an international data set. *Neurology*, 70, 868-875. doi: 10.1212/01.wnl.0000304752.50773.ec
- Didden, R., Korzilius, H., Smeets, E., Green, V. A., Lang, R., Lancioni, G. E., & Curfs, L. M. (2010). Communication in individuals with Rett syndrome: An assessment of forms and functions. *Journal of Developmental and Physical Disabilities*, 22, 105-118. doi: 10.1007/s10882-009-9168-2
- Duker, P. C., van Driel, S., van de Bercken, J. (2002) Communication profiles of individuals with Down's syndrome, Angelman syndrome and pervasive developmental disorder. *Journal of Intellectual Disability Research*, 46(1), 35-40. doi: 10.1046/j.1365-2788.2002.00355.x





References

- Hagberg, B. (1995). Rett syndrome: Clinical peculiarities and biological mysteries. *Acta Paediatrica*, 84, 971-976.
- Maes, B., Vos, P., & Penne, A. (2010). Analysis of daytime activities for children with profound intellectual and multiple disabilities in specific settings. *British Journal of Developmental Disabilities*, 56 Part 2(111), 123-136.
- Neul, J., Kaufmann, W., Glaze, D., Christodoulou, J., Clarke, A., Bahi-Biusson, N., . . . Percy, A. (2010). Rett syndrome: revised diagnostic criteria and nomenclature. *Annals of Neurology*, 68(6), 944-950. doi: 10.1002/ana.22124
- Urbanowicz, A., Downs, J., Girdler, S., Ciccone, N., & Leonard, H. (2014) Aspects of speech-language abilities are influenced by MECP2 mutation type in girls with Rett syndrome. *American Journal of Medical Genetics: Part A*, 9999, 1-9: doi:10.1002/ajmg.a.36871
- Wetherby, A. M., & Prizant, B. (2002). Communication and Symbolic Behavior Scales Developmental Profile Infant-Toddler Checklist. Baltimore, MD: Paul H. Brookes Publishing Co., Inc.

