Choice making in Rett syndrome: A descriptive study using video data

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Background

Diagnostic criteria for Rett syndrome

Main criteria	Supporting criteria

• A period of regression:
  • Loss of purposeful hand skills
  • Loss of spoken language
• Gait abnormalities
• Stereotypic hand movements
	• 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)

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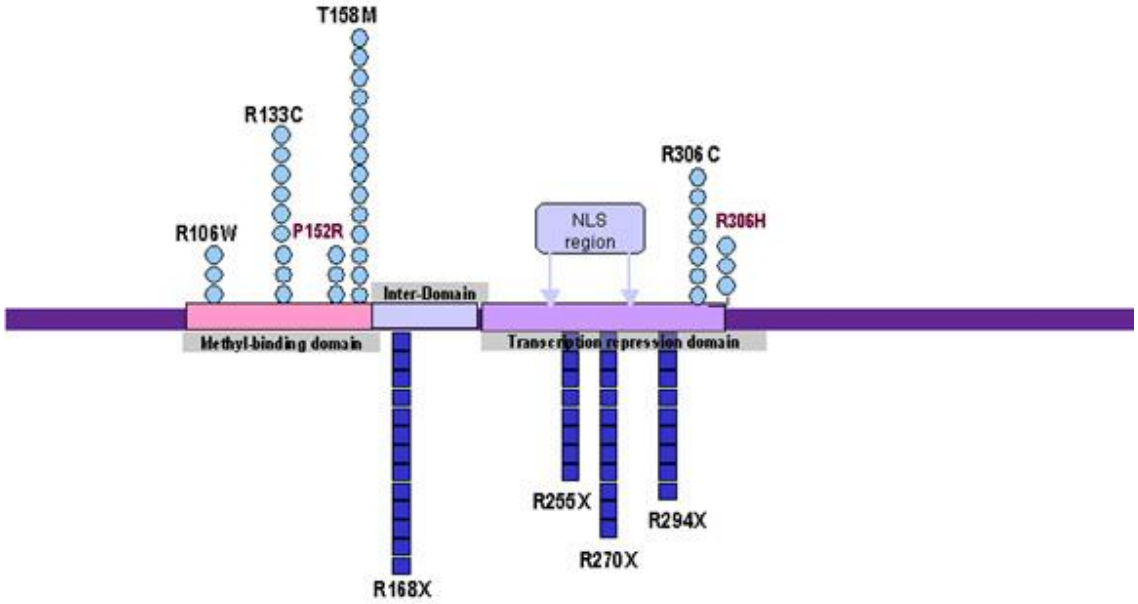
Cause of Rett syndrome



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

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Choice making in Rett syndrome

- Influences participation in everyday life (Walker, Crawford & Leonard, 2014)
- Most commonly targeted communicative function for intervention (Wandin, Lindberg, & Sonnander, 2015)
- Most common reason for using eye gaze technology (Townend et al., 2015)
- 51% - 67% females are able to make a choice (Cass et al., 2003 & Cianfaglione et al., 2015)

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What we wanted to know

- How many females with Rett syndrome can make choices?
  - How do they do this?
  - How long does it take?

- Do genotype, age, walking & grasping abilities influence choice making?

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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

Inclusion criteria

- Video clip of choice-making of females with a pathogenic *MECP2* mutation
- Videos were included if:
  - Female instructed by a communication partner to make a choice between 2 or more items
  - The items were visible
  - Video was of satisfactory quality
- If a female had more than 1 video meeting the inclusion criteria, the best video was included

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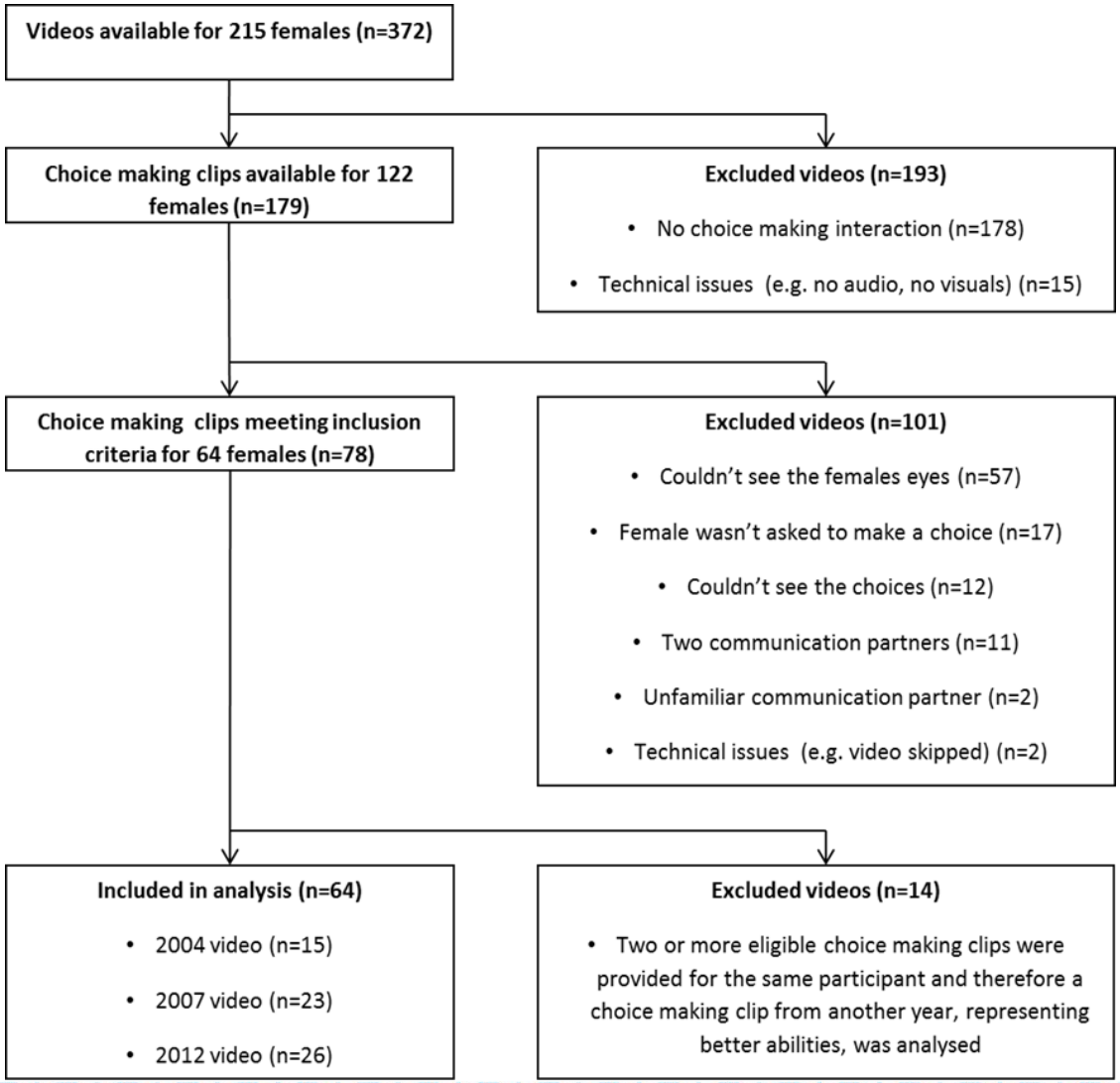
Flowchart of the selection of videos for inclusion



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Video coding

- Location of interaction
- Communication partner?
- Choice making items
- Choice made?
- Communication modalities
- Length of time taken to make choice

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Video example

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Independent variables

- Genotype
- Age groups
  - < 8 years
  - 8 < 13 years
  - 13 < 19 years
  - > 19 years
- Walking and grasping abilities

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Data analysis

- Fisher's exact test
  - Compare proportion able to make a choice or not with age group, genotype, the ability to walk & grasp objects & speech-language ability at enrolment

- Kaplan-Meier method
  - Probability of making a choice, overall & by age group

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Results

Sample characteristics

- Age range 2.3 – 35.6 years
- 82.8% made a choice
- Length in time to make choice:
  - Range 1 second to 4 minutes 6 seconds
- 72% videos filmed at home & in 87% mum was the communication partner

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Modalities used to make a choice

Modality	Frequency
Eye gaze	51
Body movements	
Takes item	7
Leans towards item	4
Gestures	
Gives item to communication partner	1
Points at item	2
Touches item without taking	7
Early sounds	2
Language	2

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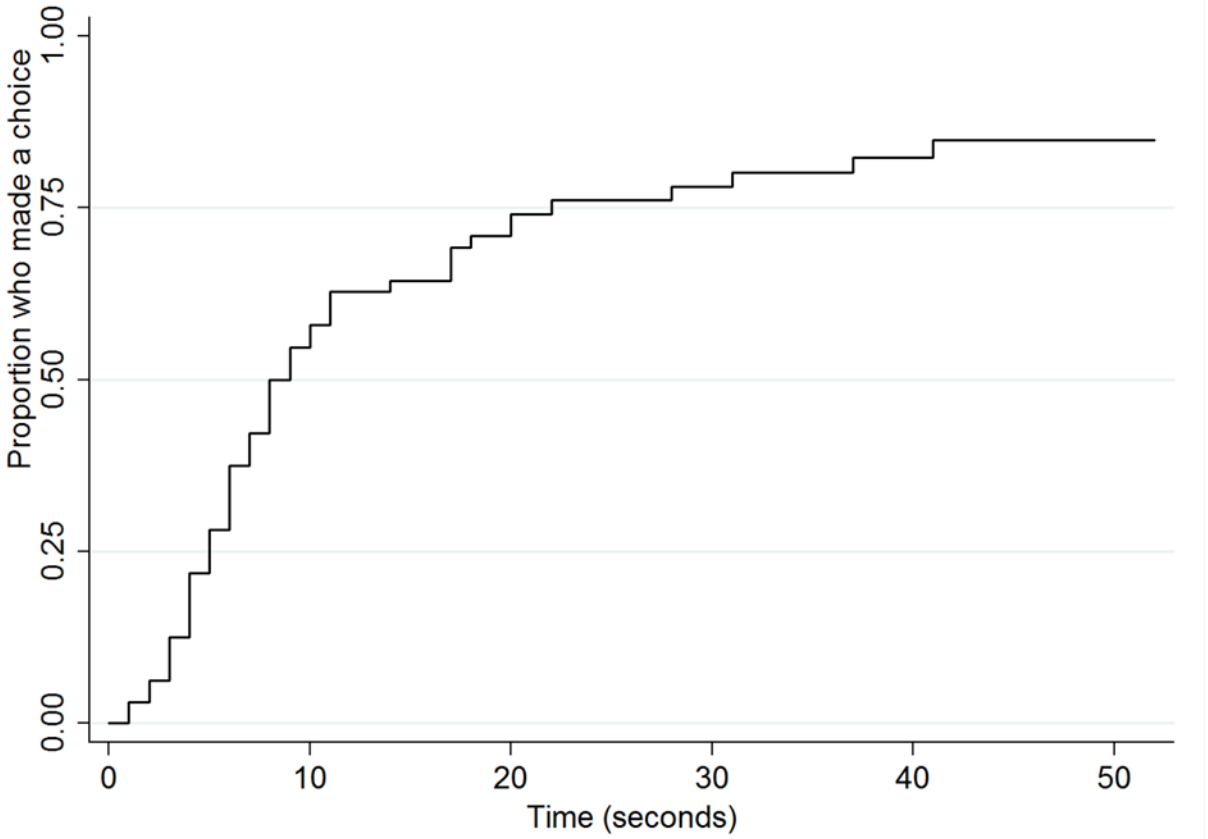
Proportion able to make a choice by age and ability to walk & grasp objects

Characteristic (n)	Able to make a choice n (%)		*p*-value
	Yes	No	
Age group (64)			
≤ 8 years (16)	14 (87.50%)	2 (12.50%)	
8 < 13 years (20)	15 (75.00%)	5 (25.00%)	
13 < 19 years (14)	13 (92.86%)	1 (7.14%)	
≥ 19 years (14)	11 (78.57%)	3 (21.43%)	0.54
Ability to walk (62)			
Independent (32)	26 (81.25%)	6 (18.75%)	
Minimal or Moderate Assistance (13)	11 (84.61%)	2 (15.38%)	
Maximal assistance or unable to walk (17)	14 (82.35%)	3 (17.65%)	1.00
Ability to grasp (58)			
Independent (33)	28 (84.85%)	5 (15.15%)	
Unable to grasp (25)	21 (84.00%)	4 (16.00%)	1.00

Proportion able to make a choice by *MECP2* mutation type

Mutation type(n)	Able to make a choice n (%)		p-value[a]
	Yes	No	
p.Arg106Trp (3)	3 (100.00%)	0	
p.Arg133Cys (6)	5 (83.33%)	1 (16.67%)	
p.Arg168* (6)	5 (83.33%)	1 (16.67%)	
p.Arg255* (6)	4 (66.67%)	2 (33.33%)	
p.Arg270* (9)	7 (77.78%)	2 (22.22%)	
p.Arg294* (6)	5 (83.33%)	1 (16.67%)	
p.Arg306Cys (5)	4 (80.00%)	1 (20.00%)	
p.Thr158Met (4)	4 (100.00%)	0	
C-terminal deletion (6)	5 (83.33%)	1 (16.67%)	
Early truncation (1)	1 (100.00%)	0	
Large deletion (4)	3 (75.00%)	1 (25.00%)	
Other (8)	7 (87.50%)	1 (12.50%)	1.00

Ability to make a choice by time



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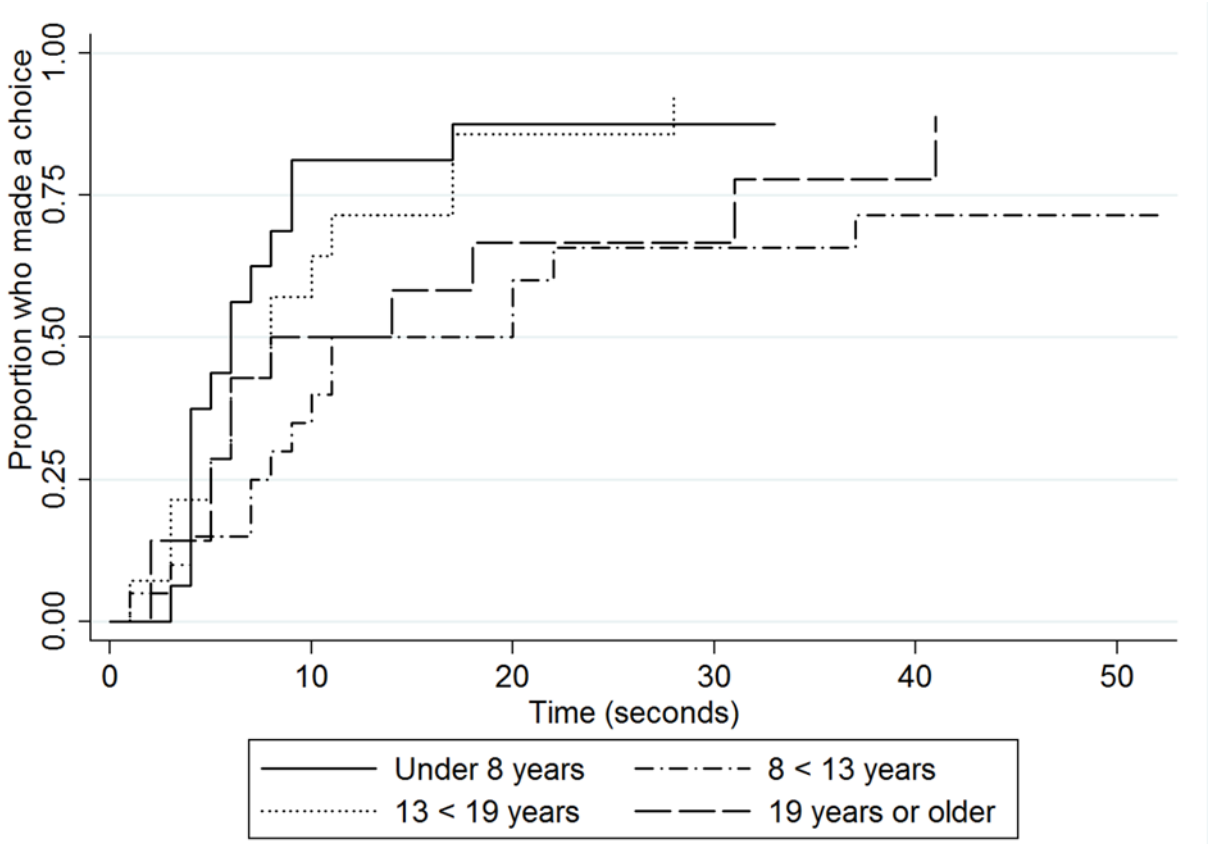
Ability to make a choice at different ages, by time



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Conclusions

Summary

- Majority were able to make a choice
- Time to make a choice varied greatly
- About 50% used a combination of communication modalities, of which eye gaze was most frequently used

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What next?

- Communication guidelines project

- Aim: To develop international clinical guidelines for the assessment, intervention and long-term management of communication in individuals with Rett syndrome, by combining available evidence with expert opinion

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