

# VCFS/22q11DS Research Laboratory at Newcastle University

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# Some of our recent research

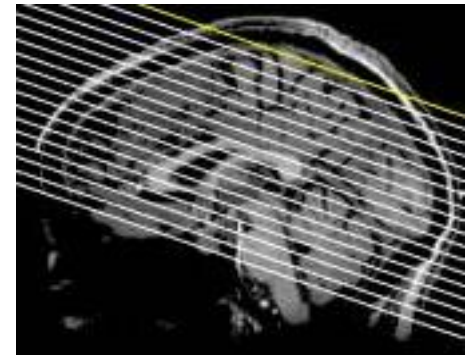
- Brain structure
- Social cognition – theory of mind and visual scanning
- Visual perception
- Coping skills and mood disorders

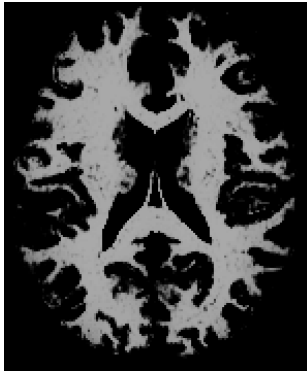
# Brain structure



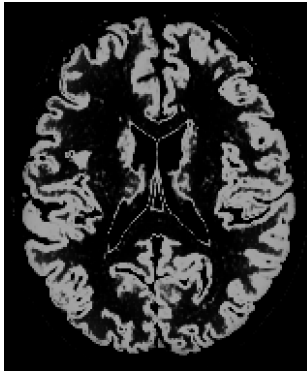
# Magnetic Resonance Imaging

- **Why use MRI?**
  - There is no dangerous radiation, it is non-invasive and harmless
  - We know there are genes in the deleted region (22q.11.2) important for brain development
  - Many cognitive processes have been linked with specific brain structures/regions
  - Many psychiatric illnesses are associated with changes of brain anatomy





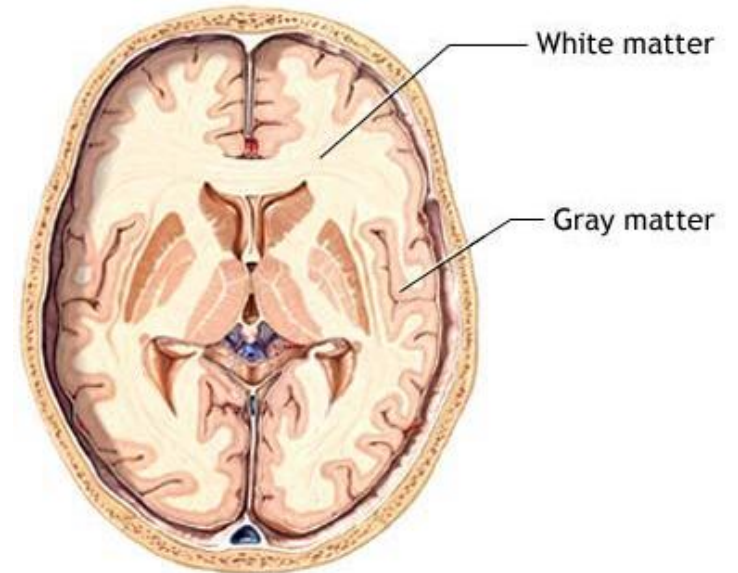
? White matter - the part of the brain and spinal cord responsible for information transmissions



? Grey matter - responsible for information processing



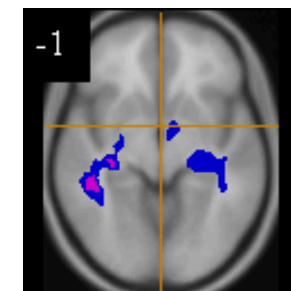
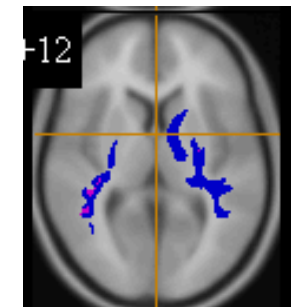
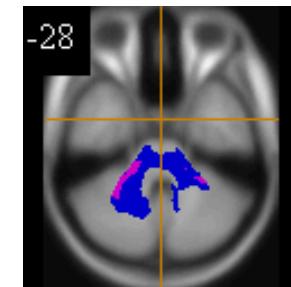
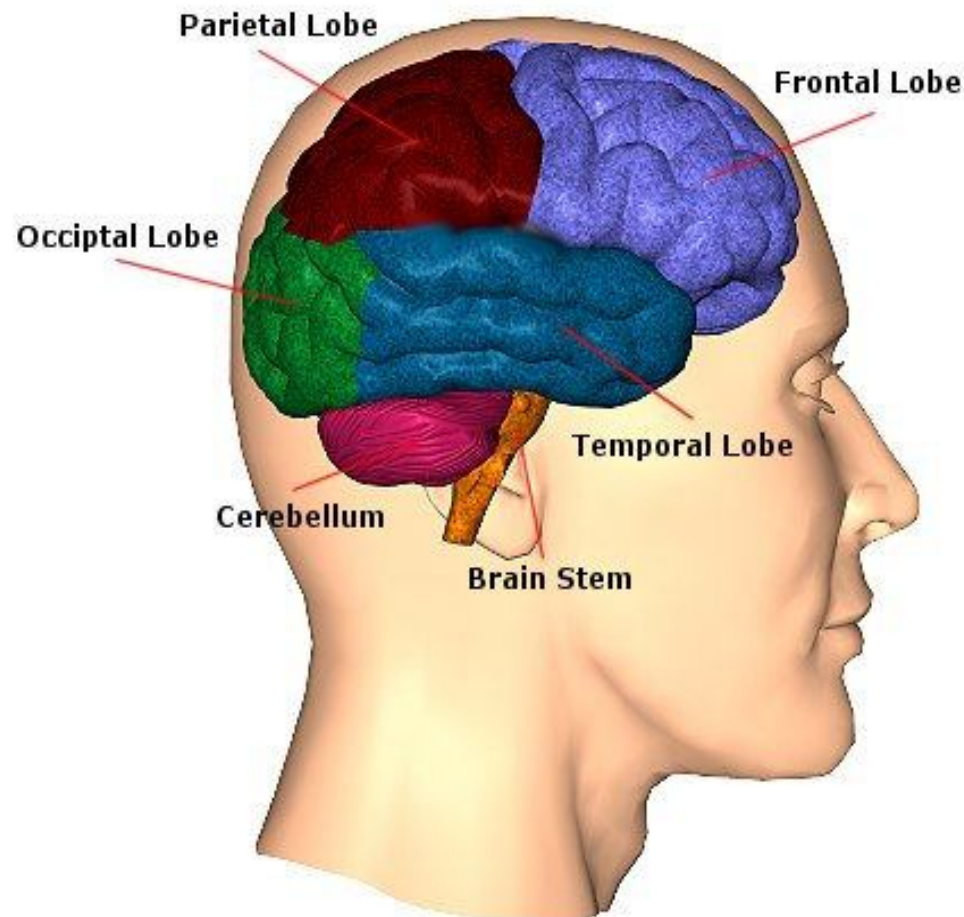
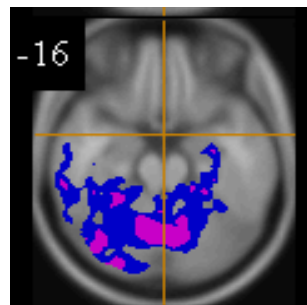
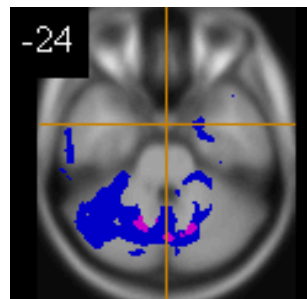
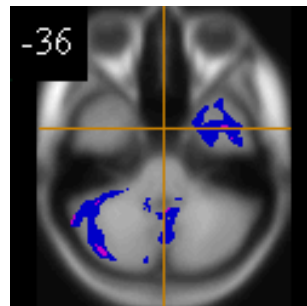
? CSF - the fluid in and around the brain, produced in the ventricles



# What have we found so far

Grey matter

White matter

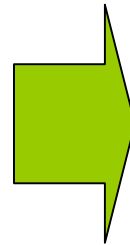


# Social cognition



# Behavioural phenotype of kids with VCFS/22q11DS

Genetic vulnerability  
Early temperament (fearful,  
separation anxiety)  
Traumatic experiences  
(hospitalisations)  
Speech problems  
Hypotonia (facial affect)  
Facial dysmorphology  
**Cognitive risk factors**



Withdrawn/socially isolated  
Extremes of behaviour incl.  
disinhibition,  
impulsiveness and shyness  
Difficulties initiating social  
interactions  
Limited range of facial  
expressions  
Social immaturity  
Anxious & fearful (phobias)  
Difficulties with cooperation,  
assertion and  
responsibility

# Theory of Mind (ToM)


- The ability to understand that other people have mental states that are independent from ones own, such as independent thoughts, beliefs and intentions.
- 2nd order: "Chris believes that Lisa wants what Ann thinks..."



# Results

VCFS (ageband \* 1st order ToM)<sup>a</sup>


Count

		Sally Ann plus smarties		Total
		fail	pass	
ageband	6-9 year	 5	23	28
	10-16 year	0	22	22
Total		5	45	50

a.

VCFS (ageband \* 2nd order ToM) <sup>a</sup>

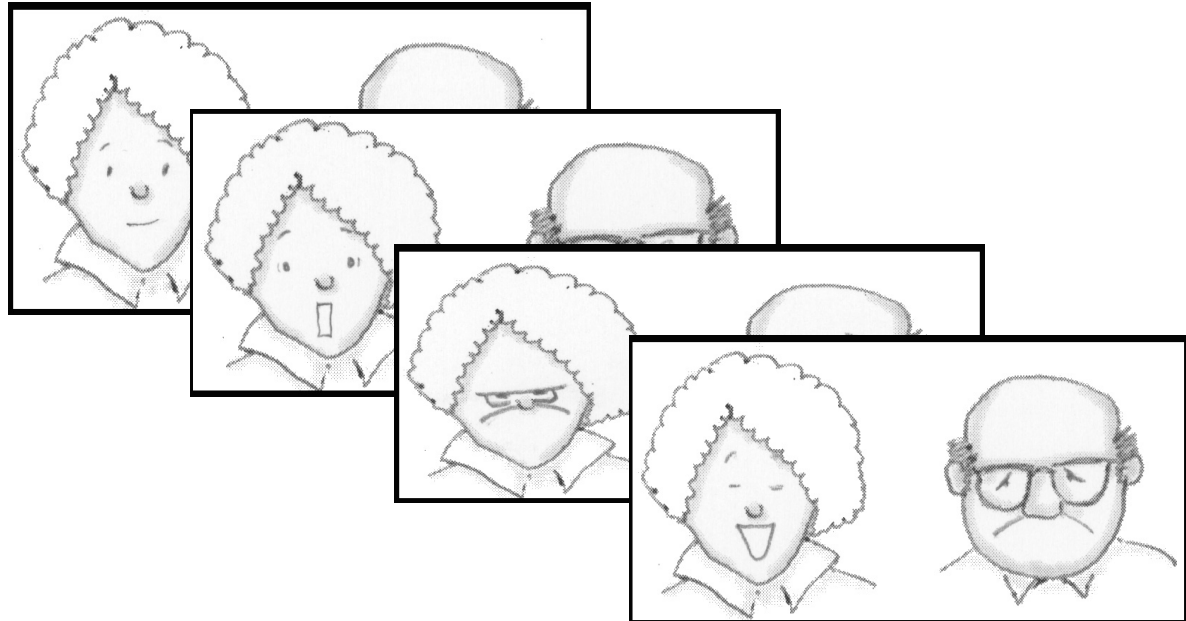
Count

		2nd order theory of mind		Total
		fail	pass	
ageband	6-9 year	 13	15	28
	10-16 year	0	22	22
Total		13	37	50

a.

Some children with VCFS/22q11DS may have a delay in understanding Theory of Mind

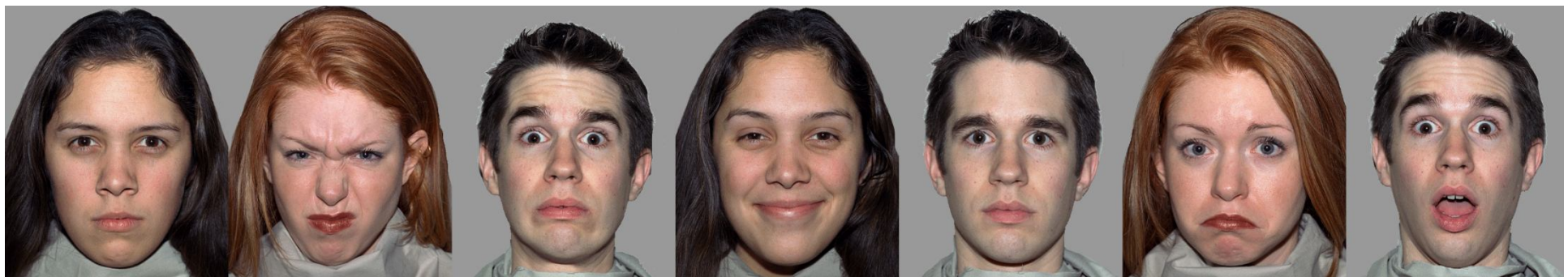
# Emotion recognition



- Children with VCFS/22q11DS also have problems with interpreting facial emotions.

# Visual Scanning of Faces Task

- Real time measure of cognitive processes
- 35 colour pictures
- neutral, happy, sad, surprise, angry, fear and disgust
- Accuracy for affect recognition



# Visual scanpaths

The VCFS/22q11DS group made **less fixations**

They also spent **more time looking outside** the face region

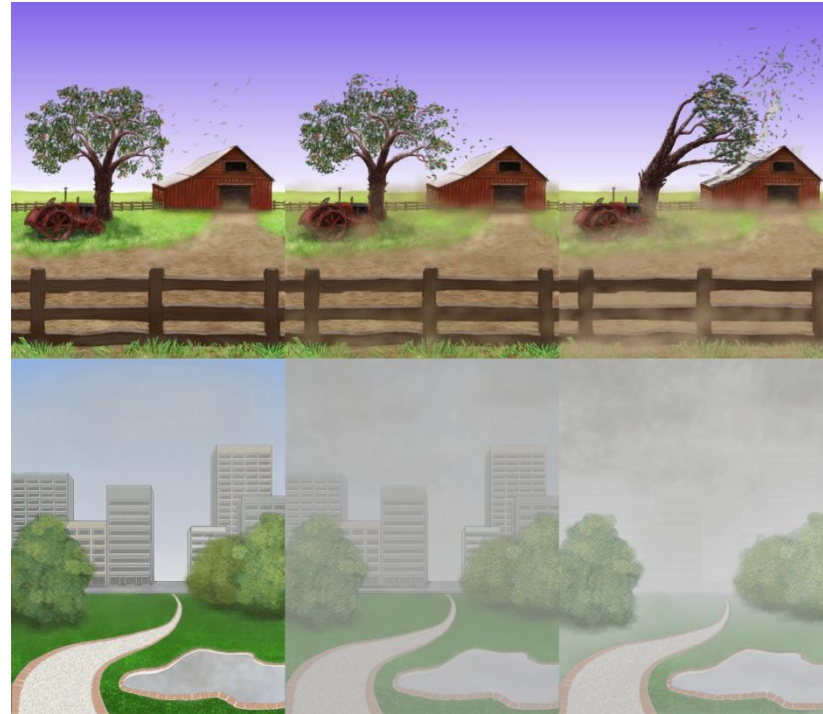
When looking at the face they spent **less time looking at salient features** of the face

It was also found that the VCFS/22q11DS groups spent significantly **less time looking at the eyes** and **longer time looking at the mouth**





- Differences in processing other images too



And smooth pursuit?



# Coping skills

**off the mark**.com by Mark Parisi

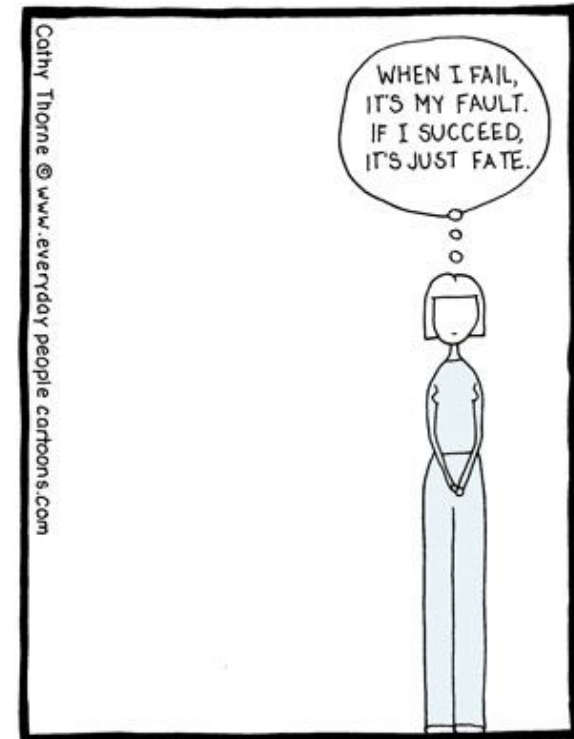


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- Coping refers to the thoughts and actions we use to deal with stress
  - Talking with friends and family
  - Trying to see things positively
  - Using humour
  - Getting emotional or instrumental support
  - Thinking about strategies to deal with stressful situations
  - Finding comfort in other activities
- Important role in 'promoting good mental health'

## Sometimes we don't use these strategies...

- Expressing negative emotions (venting)
  - Denying the specific event or emotion
  - Substance use
  - Avoiding thinking about it
  - Self-blame
- Referred to as “Maladaptive coping skills”
- Maladaptive coping skills are more common in people with anxiety/depression than in the general population



LOW SELF-ESTEEM THOUGHT #1683.

## Prevalence of Anxiety and Depression in young adults

	<i>Anxiety</i>	<i>Depression</i>	<i>Psychosis</i>
<i>General Population</i>	10%	9.7%	1%
<i>ID</i>	27%	1.3 – 40%	3%
<b><i>VCFS</i></b>	<b>60%</b>	<b>16%</b>	<b>25-30%</b>

# Other risk factors for anxiety and depression

## Individual factors

- Social Functioning
- Independence vs. Dependency
- Self-Perceived Efficacy
  - How good am I at bouncing back after bad experiences?
  - How well do I learn from mistakes?



## Rationale for studying coping in VCFS

- Little research has focused on amenable risk factors for anxiety/depression in VCFS
- One specific amenable risk factor → maladaptive coping
- High levels of anxiety and depression in VCFS adolescents, does this mean higher levels of maladaptive coping?
- By identifying such specific risk factors, you can design early targeted interventions to reduce the likelihood of anxiety/depression in VCFS

## Aims & Research Questions

***To investigate coping mechanisms and the relationship to anxiety/depression in VCFS***

Is maladaptive coping more common in VCFS than in the control group?

Is there a relationship between coping strategies and anxiety/depression?

Is the relationship between maladaptive coping and anxiety/depression mediated by individual factors (social functioning / dependence /self efficacy)?

# Methodology

## Posted questionnaires and telephone interviews with young adults and with parents

*19 young adults with VCFS/22q11DS and 16 controls aged between 15 and 24*

1. Brief COPE (Carver, 1997)

2. Social Functioning Scale (Birchwood et al, 1990)

3. Self-Perceived Efficacy Scale (Furstenberg et al, 1999)

4. Glasgow Anxiety Scale for People with an Intellectual Disability (Cuthill et al, 2003)

5. Glasgow Depression Scale for People with an Intellectual Disability (Mindham & Espie, 2001)

# Preliminary results

	Group	Mean	SD	t	p
Anxiety	VCFS	18.4	9.7	2.71	.012
	Control	9.8	5.5		
Depression	VCFS	9.4	5.8	3.24	.003
	Control	3.6	2.3		
Maladaptive coping	VCFS	20.9	5.6	.25	ns
	Control	21.4	4.9		
Adaptive coping	VCFS	37.8	6.9	1.28	ns
	Control	40.9	7.0		

*(Not yet published so we reserve the right to up-dates)*

# Preliminary results

	Group	Mean	Sd	t	p
Social Functioning	VCFS	14.75	4.62	-3.42	.005*
	Control	19.36	.67		
Dependency	VCFS	11.17	1.75	3.53	.002*
	Control	8.55	1.81		
Self-perceived Efficacy	VCFS	15.08	3.29	-4.68	.0005*
	Control	20.09	1.64		

*(Not yet published so we reserve the right to up-dates)*

# Preliminary results

**There was a significant correlation between maladaptive coping styles and anxiety in the VCFS group only**

**There was a significant correlation between maladaptive coping styles and depression in the VCFS group only**

**Individual factors did not mediate the relationship between maladaptive coping and anxiety or depression**



# Final thoughts

Maladaptive is not more common among young adults with VCFS. However, when a person with VCFS have maladaptive coping skills It is more often accompanied by depression and/or anxiety.

Therefore, our research suggests that it may be useful to improve maladaptive coping mechanisms, through targeted interventions such a cognitive-behavioural therapy, to reduce the risk of anxiety/depression.

However, the efficacy of interventions to reduce maladaptive coping in VCFS needs to be evaluated.

# What now?

We have learnt a lot, but have lots more to learn!

However, we cannot do it without your help.

Without participants in our studies we will not be able to any research.

If you want to participate in our research or want us to tell you about new research: E-mail me or contact the VCFS & 22q11 Foundation and they can forward your details to me

[Linda.e.campbell@newcastle.edu.au](mailto:Linda.e.campbell@newcastle.edu.au)



# Velo-Cardio-Facial Syndrome Educational Foundation, Inc

An international not-for-profit organization dedicated to providing support and information to individuals who are affected by Velo-Cardio-Facial syndrome, their families, physicians and other practitioners. The Foundation is independent of -- and not affiliated with -- any particular institution.

Website <http://www.vcfsef.org>

Facebook page



# Why become a member?

1 Year Membership - \$40

Student Membership - \$20

(You may also request a waiver of dues)

- VCFSEF Webinar

- Tony Simon, Kathleen Angkustsiri and Ingrid Leckliter from the M.I.N.D. Institute in California will present our first webinar on Tuesday, 20<sup>th</sup> September from 5:00-6:30 (Pacific Standard Time)

- ***Cognition, Attention and Anxiety: Implications for Everyday Functioning for individuals with VCFS/22q11.2 Deletion Syndrome***

- Powerpoint presentations from meetings

Website <http://www.vcfsef.org>



# Factsheets now available!

- Being a parent and having VCFS (Mrs Karen Ruckman)
- Being a parent of a child with VCFS (Mrs Ann Lawlor)
- Genetic Counselling (Prof Vandana Shashi)
- Cognition and Learning (Prof Ann Swillen)
- Dealing with behavioral issues at school (Dr Kevin Antshel)
- Psychiatric Illness among adults (Prof Anne Bassett)
- Brain Anatomy (Prof Wendy Kates/Dr Linda Campbell)
- Social skills (Dr Bronwyn Glaser)
- Education (Mrs Donna Landsman)

Website <http://www.vcfsef.org>