



Introduction

- Chromosome 22q11.2 Deletion Syndrome (DS22q11.2) results from a microdeletion on the 22nd chromosome and is estimated to occur in 1 out of every 4000 live births.
- Typical manifestations include heart defects, facial dysmorphisms, cleft palate, and anomalous cognitive and brain development.
 - Common cognitive phenotype includes mild to moderate reductions in IQ and impairments in numerical and visual spatial abilities
 - Neural substrate changes include overall reductions in brain volume (both gray and white matter) which appear particularly concentrated in posterior regions.
- Previous research in both typically developing children and children with neurodevelopmental disorders has suggested that visual spatial working memory is related to enumeration (counting) abilities.
- Thus, we hypothesized that the numerical impairments characteristic of 22q are the cascaded effects of alterations in the frontoparietal neural network and consequent dysfunction in spatial, attentional, and executive processing (Simon et al., 2005).
- To examine this hypothesis, we examined both visual spatial and enumeration performance in 7- to 14-year-old children with DS22q11.2 and typically developing controls.

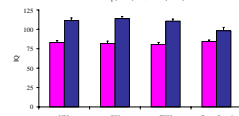
Participants

68 children between 7 & 14 years of age

(Mean = 10 years, 3 months, SD = 2 years, 2 months)

42 children with DS22q11.2 (25 female, 17 male)

26 typically developing children (12 female, 14 male)



Method

Participants completed 2 laboratory tasks:

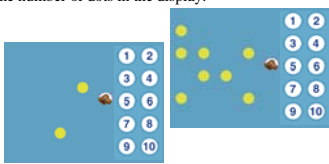
Enumeration Task on a Touchscreen Computer

Instructions: 1) Touch & count each dot on the screen. 2) Touch the dog to indicate counting complete. 3) Indicate which Arabic numeral corresponds to the number of dots in the display.

- 8 conditions:
 - Number of items (1,2,3,4,5,6,7, or 8 items)

Dependent Variables

- Reaction Time (ms)
- Percent Correct (%)
- Errors
 - Wrong Arabic Numeral or Counting Mistakes



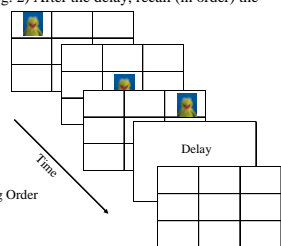
Visual Spatial Working Memory Task

Instructions: 1) Remember where you see Kermit the Frog. 2) After the delay, recall (in order) the locations in which you saw Kermit appear.

- 8 conditions:
 - Number of items (2,3,4, or 5 items)
 - Delay over which items were to be remembered
 - "Short" 500ms vs. "Long" 5000ms

Dependent Variables

- Percent Correct (%)
- Errors:
 - Wrong number of Kermits, Wrong Location, or Wrong Order



Results

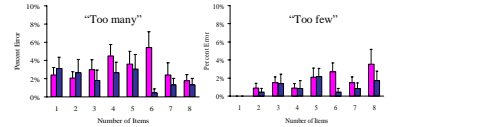
Enumeration Task on a Touchscreen Computer

Reaction Time & Percent Error

- Although both groups' reaction time was comparable across conditions, **DS22q11.2 group** made more errors (i.e., indicated the wrong number of dots were present) when counting trials with more than 2 items.
- Consistent with previous finding, **DS22q11.2** subitizing range = 2 (Simon et al., 2005)

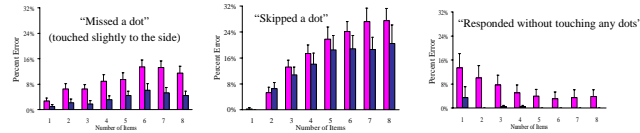
Error Types:

DS22q11.2 group had more difficulty determining the correct number of items in the display.



Why did the DS22q11.2 group make more errors?

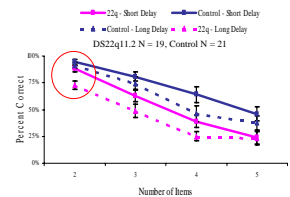
DS22q11.2 group had more difficulty producing an effective search among items in the display.



Visual Spatial Working Memory Task

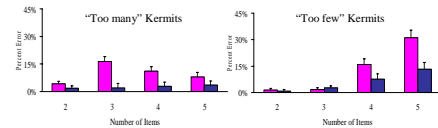
Percent Correct

- All participants' performance decreased as a function of longer delays and more items.
- DS22q11.2 group** performed similarly to the **control group** on 2-item trials at the short (500ms) delay, but below the **control group** at longer delays (5000ms).

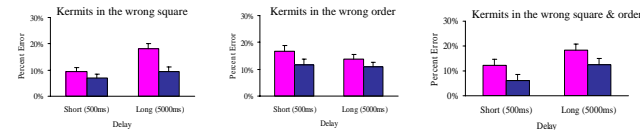


Error Types:

DS22q11.2 group had more difficulty recalling the correct locations in the sequence.



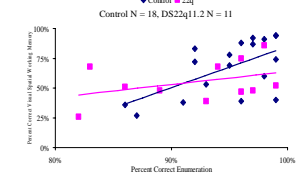
DS22q11.2 group had more difficulty recalling the correct locations and order of the sequence.



Results

Associations between tasks

- There were positive correlations between performance on the enumeration task and visual spatial working memory task across the two groups, $r(29) = .52, p < .01$.



Discussion

- Performance on the **Enumeration Task** varied between the two groups:
 - Although there was no difference in reaction time, the DS22q11.2 group made significantly more errors than the control group on items *in the counting range*.
 - This difference in performance was due to poor control of the search process in the DS22q11.2 group, likely the result of frontoparietal network impairments.
- Performance on the **Visual Spatial Working Memory Task** differed between the two groups:
 - There was no group difference in performance on 2-item trials at the short delay.
 - However, there was a group difference in performance on 2-item trials at the long delay.
 - This was due to the greater number of location errors in the DS22q11.2 group, or "poor control" of spatial location information during rehearsal.
- Combined results from these tasks offer new insights into the nature of cognitive impairments in children with DS22q11.2.

Comparisons suggest that errors made during enumeration in the DS22q11.2 group may be the result of impairments in the mental representation of and control over spatial components of display and search amongst them.

- Enumeration Task showed poor location representation which may lead to poor search patterns.
- Visual Spatial Working Memory Task showed poor location memory & poor sequence memory.
- Both tasks reveal "reduced capacity" for correctly representing spatial information.
- "Limited" or "poor" spatial representations in children with DS22q11.2 can lead to impairments in performance on cognitive tasks which require spatial planning &/or rehearsal.
- Future analyses will examine the association between error types across tasks & volumetric measurements of specific regions of interest in the brain (e.g., frontal/parietal lobes).

References

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- Shprintzen, R. J. (2005). Velo-cardio-facial syndrome. *Progress in Pediatric Cardiology*, 20(2), 187-193.
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