



## Introduction

- Males with Klinefelter Syndrome (KS) (47, XXY) and females with Trisomy X (47, XXX) have X/Y Chromosome variations in which they have an additional X chromosome.
- Research has shown that inheritance of an additional X chromosome can have deleterious effects on cognitive functioning (Bender, Linden, & Robinson, 1991; Boada, Janusz, Hutaff-Lee, & Tartaglia, 2009; Hong & Reiss, 2014).
- In particular, executive functioning (EF) is often impaired in these individuals (Lee et al., 2011, 2015; Van Rijn, Aleman, De Sonneville, & Swaab, 2009).
- Lee et al. (2015) suggested that individuals with an additional X chromosome demonstrate greater challenges with "hot" ("affectively-heavy") than "cool" ("cognitively-related") executive functioning abilities.
- Therefore, the current study 1) examined if a similar profile of EF problems is found in our sample of boys with KS (47,XXY) and girls with Trisomy X (47,XXX), and 2) compared parent ratings of EF across two parent rating scales (BRIEF/BRIEF-2 and BASC-2/BASC-3).

## Methods

### Participants

- 27 children and young adults diagnosed with Klinefelter Syndrome (N=20, all males) and Trisomy X (N=7, all females)
- Mean age = 10.6 years, SD = 4 years, range 4.7 to 20.9 years
- Mean FSIQ = 83.5, SD = 14.8, range 40-108
- All participants were evaluated by a licensed clinical neuropsychologist at Children's Healthcare of Atlanta as part of routine clinical care to assess neurocognitive strengths and weaknesses and to assist with treatment planning.

### Measures

#### Behavior Rating Inventory of Executive Function (BRIEF/BRIEF-2)

- Parent rating measure of executive function and self-regulation skills in day-to-day settings for children ages 5-18
- Includes 86 (BRIEF) or 63 (BRIEF-2) three-choice response items

#### Behavior Assessment System for Children (BASC-2/BASC-3)

- Parent rating of emotional and behavioral functioning
- There are three age levels (2-5, 6-11 and 12-21 years) that contain 139-175 four-choice response items
- Analyses utilized two Content scales of the BASC, those assessing Emotional Control and Executive Functioning.
- Due to small N the KS and Trisomy X were combined for analyses

## Results

**Table 1. BRIEF/BRIEF-2 and BASC-2/BASC-3 Scores**

	N	Min	Max	M	SD
BRIEF					
Inhibit	16	40	81	61.4	14.6
Shift	16	40	91	63.6	16.3
Emotional Control	16	37	85	63.6	16.0
Behavioral Regulation Index (BRI)	15	38	88	63.9	15.8
Initiate	15	41	82	62.3	13.7
Working Memory	16	40	98	67.2	16.5
Plan/Organize	16	37	79	61.1	12.6
Organization of Materials	15	41	72	56.3	10.5
Monitor	15	34	82	61.5	13.6
Metacognitive Index (MI)	16	41	89	64.6	15.0
Global Executive Composite (GEC)	16	40	89	66.1	15.6
BRIEF-2					
Inhibit	11	45	75	60.5	9.5
Self-Monitor	11	44	78	63.1	9.7
Behavioral Regulation Index (BRI)	11	45	76	62.4	8.8
Shift	11	43	86	68.4	14.8
Emotional Control	11	51	88	68.0	11.3
Emotional Regulation Index (ERI)	11	47	89	69.6	12.8
Initiate	11	42	79	63.3	10.4
Working Memory	11	52	84	66.6	11.1
Plan/Organize	11	45	80	64.2	11.6
Task-Monitor	11	35	73	59.6	11.3
Organization of Materials	11	45	73	54.3	9.2
Cognitive Regulation Index (CRI)	11	48	79	63.3	10.5
Global Executive Composite (GEC)	11	47	84	67.2	9.4
BASC-2/3					
Emotional Control	25	37	94	60.8	13.1
Executive Functioning	25	34	84	61.4	11.3

Note. T-scores are reported for each BRIEF/BRIEF-2 and BASC-2/3 subscale (M = 50, SD = 10). For the BRIEF, T-scores of 65-69 are considered potentially clinically elevated and T-scores above 70 are considered clinically elevated. For the BASC, T-scores 60-69 are considered at risk, and T-scores 70 and above are considered clinically elevated.

**Table 2. Correlations between BRIEF and BASC**

	N	BASC ESC	BASC EF
BRIEF subscales			
Inhibit	25	0.58**	0.66**
Shift	25	0.64**	0.59**
Emotional Control	25	0.74**	0.60**
Behavioral Regulation Index	14	0.86**	0.84**
Initiate	25	0.56**	0.69**
Working Memory	25	0.64**	0.81**
Plan/Organize	25	0.46*	0.67**
Organization of Materials	25	0.64**	0.81**
Monitor	14	0.86**	0.84**
Metacognition Index	14	0.86**	0.91**
Global Executive Index	14	0.88**	0.91**

Note. Due to the small number of BRIEF-2's, data from overlapping subscales from the BRIEF and BRIEF-2 were combined and are reported here. BASC ESC = BASC Emotional Self-Control content scale. BASC EF = BASC Executive Functioning content scale. \*Correlation is significant at the 0.05 level (2-tailed). \*\*Correlation is significant at the 0.01 level (2-tailed).

## Results

- Analyses revealed significant, moderate to large correlations between the two BASC content scales related to EF skills, and all BRIEF scales.
- The BRIEF ratings indicated slightly greater difficulties with emotional control and shifting than other areas of behavioral and emotional regulation, and slightly greater difficulties with working memory than other areas of cognitive regulation.
- Significantly fewer problems were reported with Organization of Materials than Shift, Emotional Control, Initiate, Working Memory, and Plan/Organize,  $p = .03, .02, .04, <.001, \text{ and } .03$ .
- Lack of other significant findings may be due in part to small sample size, or to individual variability in the functioning level of individuals with these conditions.

## Conclusions

- Results suggest the presence of an additional X chromosome (in KS and Trisomy X) is associated with a relatively homogenous EF profile but with some relative strengths and weaknesses.
- Although BRIEF index scores were not significantly different, children in this sample appeared to have slightly more difficulty with emotional regulation than cognitive regulation skills, consistent with prior researching indicating more difficulties with "hot" than "cold" executive functioning skills in this population.
- The strong correspondence between BASC executive function content scales and BRIEF ratings of executive function suggests that these BASC content scales could provide an alternate way of examining EF in this population.

## References

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