PHONOLOGICAL WORKING MEMORY ON A NONWORD REPETITION TASK IN INDIVIDUALS WITH GALACTOSEMIA

By

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PHONOLOGICAL WORKING MEMORY ON A NONWORD REPETITION TASK IN
INDIVIDUALS WITH GALACTOSEMIA

Abstract
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Introduction

Classic galactosemia is a recessive inborn error of metabolism that results in speech or
language disorders in 60-80% of affected individuals. Nonword repetition tasks have been used
extensively to assess phonological working memory of individuals with language impairments.
The repetition of nonwords has been proposed to be relatively independent of level of cognitive
function. A nonword repetition task (NRT) was used to determine whether individuals with
galactosemia (IWG) differ from healthy controls (HC) in phonological working memory. A
relation between performance on the NRT and full scale IQ (FSIQ) scores in IWG was assessed.
Finally, evidence for neurodegeneration in phonological working memory across ages of IWG
was investigated.

Method

The NRT was administered to 162 children aged 4;0-16;11 and 43 adults aged 17;0-69;11. Thirty-two of the children and 33 adults had been diagnosed with galactosemia. All
participants were administered the Goldman-Fristoe Test of Articulation-2 (GFTA-2). In
addition, the galactosemia group was administered the Peabody Picture Vocabulary Test-4
(PPVT-4), the Oral and Written Language Scale (OWLS) and a full scale IQ (FSIQ) test.
Results

Results indicated significantly lower scores on the NRT for IWG than for HC groups. Relationships were found among NRT scores and FSIQ, increasing age, articulation, and receptive language, and the presence of childhood apraxia of speech (CAS).

Discussion

IWG have deficits in phonological working memory compared to HC as measured by performance on the NRT. Performance on the NRT provided no evidence of neurodegeneration with increasing age for IWG.
# TABLE OF CONTENTS

<table>
<thead>
<tr>
<th>Section</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>SIGNATURE PAGE</td>
<td>ii</td>
</tr>
<tr>
<td>ABSTRACT</td>
<td>iii</td>
</tr>
<tr>
<td>LIST OF FIGURES</td>
<td>vii</td>
</tr>
<tr>
<td>LIST OF TABLES</td>
<td>viii</td>
</tr>
<tr>
<td>CHAPTER</td>
<td></td>
</tr>
<tr>
<td>1. INTRODUCTION</td>
<td>1</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>1</td>
</tr>
<tr>
<td>Neurodegeneration</td>
<td>2</td>
</tr>
<tr>
<td>Nonword repetition task</td>
<td>4</td>
</tr>
<tr>
<td>Research questions</td>
<td>5</td>
</tr>
<tr>
<td>2. METHOD</td>
<td>6</td>
</tr>
<tr>
<td>Participants</td>
<td>6</td>
</tr>
<tr>
<td>Materials and Instrumentation</td>
<td>7</td>
</tr>
<tr>
<td>Procedure</td>
<td>8</td>
</tr>
<tr>
<td>Scoring</td>
<td>8</td>
</tr>
<tr>
<td>Reliability</td>
<td>8</td>
</tr>
<tr>
<td>Data analysis</td>
<td>9</td>
</tr>
<tr>
<td>3. RESULTS</td>
<td>10</td>
</tr>
<tr>
<td>Nonword repetition task results</td>
<td>10</td>
</tr>
<tr>
<td>FSIQ results</td>
<td>11</td>
</tr>
<tr>
<td>Neurodegeneration</td>
<td>12</td>
</tr>
</tbody>
</table>
Figures..........................................................10
Tables...........................................................11
4. DISCUSSION................................................13
5. REFERENCES ................................................18
LIST OF FIGURES

1. MEAN SCORES ON NONWORD REPETITION TASK ........................................10
LIST OF TABLES

1. MEANS AND STANDARD DEVIATIONS ................................................................. 11
2. RELATIONSHIPS AMONG NRT AND VARIABLES .............................................. 12
PHONOLOGICAL WORKING MEMORY ON A NONWORD REPETITION TASK IN INDIVIDUALS WITH GALACTOSEMIA

CHAPTER I

INTRODUCTION

Introduction

Classic galactosemia is an autosomal, recessively inherited, inborn error of metabolism that occurs in about 1:60,000 babies born in the United States (Nelson et al., 1991). It causes life-threatening illness (E. coli, increased intracranial pressure with cerebral edema, and encephalopathy) within days of feeding galactose-containing milk to a newborn (Ridel, Leslie, & Gilbert, 2005). Progressive liver and kidney damage may also lead to death soon after ingesting galactose (Nelson, Waggoner, Donnell, Tuerck, & Buist, 1991). The initial symptoms resolve upon introduction of a galactose-restricted diet (Nelson et al., 1991). However, despite dietary restriction of galactose, these individuals have significant complications later in life (cataracts, ovarian failure, cognitive impairment, and apraxia of speech; Ridel et al., 2005). Galactose is present in mammalian milk in the form of disaccharide lactose. It is hydrolyzed in the intestine to the monosaccharides galactose and glucose, which are then absorbed. The enzyme galactose-1-phosphate uridylic transferase (GALT) is then responsible for converting galactose-1-phosphate (GAL-1-P) to glucose-1-phosphate. A deficiency of GALT results in toxic accumulation of GAL-1-P in the blood and tissues even in well treated cases (Ridel et al., 2005).

A possible risk factor for IWG is the homozygous Q188R genotype. Potter, Lazarus, Johnson, Steiner, and Shriberg (2008) found that CWG with the Q188R/Q188R genotype were at greater risk than CWG with Q188R/other or other/other genotypes for cognitive and language impairment. Ridel et al. (2005) stated that although 167 mutations of the GALT gene have been identified, Q188R homozygous is the most prevalent mutation in the Western population. Other
genotypes such as mutation S135L, which is more commonly present in the African-American population, and has a milder phenotype often with minimal impact on speech and language (Ridel et al., 2005).

The severity of neurologic abnormalities has been found to differ among IWG. Neurodegeneration is one potential complication for IWG, although it has been reported only in a few cases. Friedman, Levy, and Boustany (1989) proposed that as IWG age, a spectrum of neurological diseases may present, including seizures, cerebellar ataxia, extrapyramidal dysfunction (abnormal involuntary movements), and apraxia. Manis, Cohn, McBride-Chang, Wolff, and Kaufman (1997) proposed that neurodegeneration may be the exception rather than the rule for IWG. A case study by Friedman et al. (1989) observed evidence of neurodegeneration in a 46-year-old woman and her 41-year-old brother who were both diagnosed with galactosemia at birth and placed on a galactose-restricted diet. Neurological exams indicated that the woman showed Purkinje cell dropout (decrease in neurons located in the cerebellar cortex), dentate gliosis (proliferation of astrocytes in damaged areas of the central nervous system), and a tumor in her basal ganglia causing diminished arm swing, dystonic hand posture, and arm rigidity. Her severe apraxia may have represented cortical dysfunction. Jan & Wilson (1973) reported a case study involving a 25-year-old man with galactosemia who was not diagnosed until age 13. Observations showed significant decline in neurological function with age. Histopathological studies showed cerebral cortical neuronal degeneration, cerebral white matter atrophy and sclerosis, and cerebellar degeneration.

A possible mechanism to explain progressive central nervous system involvement is the inability of IWG to synthesize uridine diphosphate galactose (UDP-galactose). UDP-galactose serves as a building block for galactolipid and mucopolysaccharide synthesis, which are essential
for myelin formation (Coet, Suzuki, & Popko, 1998). Pathologically abnormal myelination is a prominent feature in IWG (Ridel et al., 2005). Ng, Xu, Kaufman, & Donnell (1989) found that UDP-galactose levels of IWG were substantially lower than in controls, and hypothesized that UDP-galactose is responsible for the late onset clinical manifestations in galactosemia (ovarian failure, speech deficit, and neurological abnormalities). It was hypothesized that uridine supplementation could improve synthesis of UDP-galactose. However, this hypothesis was later disputed by Manis et al., (1997) who supplemented 35 IWG with dietary uridine over a period of two to five years and found no support for any uridine treatment-related change.

Research has shown that galactosemia causes long-term cognitive deficits with 50% of affected IWG having borderline to low IQ (below 85) (Waggoner, Buist, & Donnell, 1990). Shield, Wadsworth, MacDonald, Stephenson, Tyfield, & Holton (2000) found that despite the restricted diet, subjects’ IQ scores remained in borderline to low ranges. In a study of 60 children with galactosemia, ages five to 15 years, the mean IQ standard score was 76 (Kromrower & Lee, 1970). Manis et al., (1995) and Ridel et al., (2005) also reported that mean IQ scores are reduced in both CWG and AWG. Potter et al. (2008) reported that CWG, particularly those with the Q188R/Q188R genotype, score lower on IQ tests. A large cross-sectional study by Wagoner, Buist & Donnell (1990) found that mean IQ scores of patients with galactosemia declined by 6.2 points from age 3-5 to age 6-9, and by 4.4 points from 6-9 to 10-16 years of age. Language impairment in CWG is related to their level of cognitive function (Potter et al., 2008). Eighty-eight percent of CWG with borderline-low FSIQ scores (84 and below), and 56% of CWG with FSIQ scores of 85 and above have language impairment (Potter et al., 2008).

Nonword repetition performance is an indicator of language impairment, as it
distinguishes between children with language impairment and language normal (LN) children with a high degree of accuracy (Dollaghan & Campbell, 1998). When investigating working memory capacity in children with specific language impairment (SLI; defined as normal IQ and language impairment), children with SLI showed poorer word recall than their normal language controls indicating significantly poorer verbal working memory (Ellis Weismer, Evans, & Hasketh, 1999). Other studies examining nonword repetition found that children with SLI and LI (defined as borderline to low IQ and language impaired) repeated nonwords less accurately than healthy controls (Bishop, North, & Donlan, 1996; Marton & Schwartz, 2003; Montgomery, 1995b). Children with SLI are also less accurate at repeating sentences, even after adjustment for decreased receptive vocabulary (Montgomery, 1995a). Although phonological working memory, as measured by nonword repetition, is a predictor of language impairment in children with SLI, there is currently no research on phonological working memory in IWG.

Previous studies examining the relationship between language and nonword repetition excluded children with co-occurring speech sound disorders (SSD). IWG are not only at risk for language impairments but are also at risk for co-occurring motor speech disorders (MSD) affecting 24-65% of IWG (Shriberg, Potter, & Strand, 2010; Webb, Singh, Kennedy, & Elsas, 2003). Presence of a speech sound disorder including MSD diagnoses of childhood apraxia of speech (CAS) and dysarthria (DYS) may affect IWG’s performance on measures of phonological working memory.

The nonword repetition task (NRT) is commonly used to assess phonological working memory (Dollaghan & Campbell, 1998). The NRT consists of 16 nonwords ranging from one to four syllables, with four words at each syllable length. The nonwords included in the NRT are composed of early-developing sounds, which minimizes errors due to an individual’s ability to
produce them. The NRT nonwords were 1) constructed to minimize familiarity and predictability by insuring that none of their individual syllables corresponded to an English word, 2) contain only tense vowels, which are longer in duration than weak syllables, and 3) differ from the typical metrical stress pattern in American English that alternates strong and weak syllables. The NRT closely matches the phonological component of word learning, and correlates with measures of phonological working memory. As the task elicits activation of many language processes, and is proposed to be relatively independent of level of cognitive function, it is a powerful tool that can be used to identify children with various language impairments (Coady & Evans, 2008).

Individuals with idiopathic language impairments typically have decreased phonological working memory. Since most IWG have language impairments, they are likely also at risk for decreased phonological working memory. Case studies have reported progressive declines in language in some IWG with increasing age. A progressive decline with increased age in IWG may be evidence for neurodegeneration. The purpose of the present study is to examine phonological working memory in IWG and to look for evidence of neurodegeneration across ages by answering the following questions:

- Do individuals with galactosemia differ from healthy controls in phonological working memory as measured by the nonword repetition task?
- Does performance on the nonword repetition task relate to IQ in individuals with galactosemia?
- Is there evidence of neurodegeneration in phonological working memory across ages of individuals with galactosemia?
CHAPTER II

METHOD

Participants

Participants in this study included 162 children and 43 adults. Thirty-two children had galactosemia and a history of speech sound disorders (SSD) and 130 were typically developing healthy controls (HC). The children with galactosemia, ages 4;0-16;11, were recruited through two parent support groups, Parents of Galactosemic Children and Galactosemic Children of Minnesota and through metabolic clinics throughout the United States and tested in their homes. Hearing was screened for all participants using puretone audiometry at 25dB for 1,000, 2,000, and 4,000Hz. All children with galactosemia had normal hearing with the exception of one child with a mild (30-35 dB) bilateral hearing loss. HC children, five boys and five girls from each of the following age groups: 4;0-4;11, 6;0-6;11, 8;0-8;11, 10;0-10;11, 12;0-12;11, 14;0-14;11, and 16;0-16;11, were recruited from and tested at preschools and public schools in Eastern Washington. All HC children were academically at grade level and had never been referred for special education services, including speech and language, as reported by parents and teachers. All HC children had normal hearing.

Thirty-three adults had galactosemia and 10 adults served as HC. The adults with galactosemia, ages 18-59 years, were recruited through Parents of Galactosemic children and tested in a conference room adjacent to Children’s Hospital Boston. Some, but not all of the adults with galactosemia had received special educational services including speech and language, by self-report. Self-reporting of special education services was compared to medical records for the adults with galactosemia who had received medical care at Children’s Hospital.
Boston. Self-report differed from hospital records for some AWG. All AWG had normal hearing except two adults with mild unilateral high frequency loss and one adult with mild bilateral high frequency hearing loss. HC adults were recruited from Eastern Washington through personal contacts and were tested at a university speech and language clinic. HC included one male and one female from the following groups: 18-19, 20-29, 30-39, 40-49, and 50-59 years. All HC adults had normal hearing. No HC adults had received special education services, including speech and language, by self-report.

The presence or absence and type of an MSD was determined using the adapted Mayo Clinic Motor Speech Disorder Classification System (Potter, 2010; Shriberg, Potter, Strand, 2010).

**Materials and Instrumentation**

Articulation skills of all participants were assessed using the Goldman-Fristoe Test of Articulation-2 (GFTA-2; Goldman & Fristoe, 2000). Full scale intelligence quotient (FSIQ) standard scores were measured using the Kaufman Brief Intelligence Test-2 (KBIT-2; Kaufman & Kaufman, 2004) for CWG, and the Wechsler Adult Intelligence Scale (WAIS; Wechsler, 2008) for AWG. Receptive and expressive language skills of the children with galactosemia were assessed with the Oral and Written Language Scale (OWLS; Carrow-Woolfolk, 1996). Receptive vocabulary skills of the adults with galactosemia were assessed with the Peabody Picture Vocabulary Test (PPVT-4; Dunn & Dunn, 2007). FSIQ and language skills of the HC children and adults were not formally assessed. Recordings for the three data sets described in the text were made in quiet environments in participant’s homes, educational facilities, and clinical research facilities. Speech samples for 15 of the CWG were recorded on a Sony DCR-DVD301 digital video recorder using a Shure WH30TOG cardioid microphone. Samples for 17 CWG, all AWG, and the HC reference database were recorded on a Marantz CDR 420 digital audio recorder and a Shure MX412D/C
microphone.

**Procedure**

All participants were given the same instructions prior to taking the NRT: “You are going to say some silly words. Listen carefully and then copy the words exactly the way she says them. Are you ready?” Nonwords were then presented one at a time via an audio recording. Immediately upon hearing each nonword, participants repeated them.

**Scoring**

Scoring of the NRT was completed by a trained graduate student who was not one of the examiners, and was blind to participants’ diagnosis and treatment status. The scoring procedures used by Dollaghan and Campbell (1998) were followed in this study. Each phoneme was scored as correct or incorrect relative to its target phoneme. Substitutions and omissions were scored as incorrect, while distortions were scored as correct. Phoneme additions did not count as errors.

In instances where the syllable structure of nonwords was not maintained (adding or omitting), phoneme scoring proceeded after aligning the syllable sequence produced by the participant as nearly as possible to that of the target. This anchoring technique was designed to maximize the participant’s score. The number of phonemes repeated correctly were divided by the total number of targets and then multiplied by 100 to obtain the Percentage of Phonemes Correct (PPC) for one syllable (1PPC), two syllables (2PPC), 3 syllables (3PPC), 4 syllables (4PPC), and for the entire list of nonwords (Total PPC). All other assessments were scored according to their individual test directions.

**Reliability**

Recordings from nine randomly selected adult participants (20%), and 33 randomly selected child participants (20%) were transcribed independently by a second trained graduate
student. Phoneme-by-phoneme percentages of agreement of scores averaged 92% for adult participants, and 93% for child participants.

Data Analysis

This cross-sectional study used an analysis of covariance (ANCOVA) to compare mean NRT scores at each syllable length for IWG to those of healthy controls. Age group was used as a partial variable. A multiple regression analysis was used to look for potential evidence of neurodegeneration across ages of individuals in the galactosemia and control groups with age group as the independent variable (IV), while the NRT score (at each syllable length) was the dependent variable (DV). A Pearson product moment correlation was used to analyze correlations among NRT scores at each syllable length, FSIQ, CAS, DYS, receptive language, and articulation.
CHAPTER III
RESULTS

In the present study the first research question addressed whether IWG differ from HC in phonological working memory as measured by the NRT. It was found that IWG scored significantly lower at each syllable length, including PPC at one syllable (F_{1, 214} = 50.88, p < .001), two syllables (F_{1, 214} = 71.56, p < .001), three syllables (F_{1, 214} = 85.04, p < .001), four syllables (F_{1, 214} = 51.43, p < .001), and total PPC (F_{1, 214} = 96.09, p < .001) as compared to healthy controls. The galactosemia and control groups scored significantly lower at the 4PPC length than PPC’s at all shorter lengths. Mean scores on the NRT for AWG and healthy control adults are presented in Figure 1. Means and standard deviations of NRT performance for PPC of all participants are presented in Table 1.

Figure 1. Mean scores on the Nonword Repetition Task (NRT) for percent phonemes correct (PPC) for adults in the galactosemia group (light grey) and control group (dark grey) for the total task and at each syllable length.
Table 1. Means and standard deviations of Nonword Repetition Task for percentage phonemes correct (PPC) for the total task and each syllable length by group.

<table>
<thead>
<tr>
<th></th>
<th>1PPC</th>
<th>2PPC</th>
<th>3PPC</th>
<th>4PPC</th>
<th>TOTPPC</th>
</tr>
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<tbody>
<tr>
<td>CWG</td>
<td>76.81 (15.85)***</td>
<td>74.1 (23.99)***</td>
<td>68.94 (23.09)***</td>
<td>56.58 (25.58)***</td>
<td>62.42 (21.69)***</td>
</tr>
<tr>
<td>AWG</td>
<td>84.56 (12.9)*</td>
<td>91.91 (10.28)</td>
<td>82.67 (18.15)*</td>
<td>67.97 (21.55)*</td>
<td>79.32 (15.54)*</td>
</tr>
<tr>
<td>HC-C</td>
<td>91.09 (9.38)***</td>
<td>95.12 (7.19)***</td>
<td>89.87 (9.4)***</td>
<td>77.33 (13.27)***</td>
<td>85.98 (9.4)***</td>
</tr>
<tr>
<td>HC-A</td>
<td>94.12 (5.47)*</td>
<td>98 (2.99)</td>
<td>97.5 (4.19)*</td>
<td>80.83 (8.59)*</td>
<td>90.94 (4.26)*</td>
</tr>
</tbody>
</table>

CWG = children with galactosemia  
AWG = adults with galactosemia  
HC-C = healthy child controls  
HC-A = healthy adult controls

* p < .05, ** p < .01, *** p < .001

As compared to healthy controls, IWG more frequently substituted real words for nonwords at the single syllable length. For example, IWG typically substituted the word “rope” for the nonword /voup/. Fifty-eight percent of CWG and 30% of AWG exhibited this pattern at the single syllable length. In contrast, 20% of children and 20% of adults in the HC groups replaced a nonword with a real word.

The second research question of the present study addressed whether performance on the NRT related to FSIQ in IWG. The mean FSIQ standard score for the CWG group was 86 (range: 57 to 111), and the mean FSIQ for AWG was 88 (range: 55 to 122). IWG with borderline to low FSIQ (84 and below; N = 29) had lower performance on the NRT than IWG with average FSIQ scores (85 and above; N = 36) at 1PPC (F<sub>1,63</sub> = 4.78, p < .05), 2PPC (F<sub>1,63</sub> = 4.76, p < .05), 3PPC (F<sub>1,63</sub> = 5.32, p < .05), 4PPC (F<sub>1,63</sub> = 9.65, p < .01), and TOTPPC (F<sub>1,63</sub> = 12.35, p < .001). Overall, IWG diagnosed with an MSD had lower NRT scores than individuals not diagnosed with an MSD. When viewed by specific MSD, the presence of CAS in IWG negatively correlated with NRT scores (r = 0.402, p < .05), indicating that IWG with CAS had poorer phonological working memories as compared to IWG not diagnosed with an MSD. IWG
diagnosed with dysarthria (DYS) did not differ from IWG not diagnosed with an MSD (p > .05). IWG and borderline to low FSIQ had poorer performance on the NRT, lower receptive language, and poorer articulation, as compared to HC groups (Table 2).

Table 2. The relationship (r values) among Nonword Repetition Task performance for percentage phonemes correct (PPC) for each syllable length and the total task and FSIQ, MSD, receptive language, and articulation.

<table>
<thead>
<tr>
<th></th>
<th>FSIQ</th>
<th>MSD</th>
<th>Receptive Language</th>
<th>Articulation</th>
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<tr>
<td>1PPC</td>
<td>0.282*</td>
<td>0.388**</td>
<td>0.386*</td>
<td>0.457***</td>
</tr>
<tr>
<td>2PPC</td>
<td>0.258*</td>
<td>0.358**</td>
<td>0.129</td>
<td>0.420***</td>
</tr>
<tr>
<td>3PPC</td>
<td>0.314*</td>
<td>0.351**</td>
<td>0.267*</td>
<td>0.327**</td>
</tr>
<tr>
<td>4PPC</td>
<td>0.394**</td>
<td>0.436***</td>
<td>0.271*</td>
<td>0.309*</td>
</tr>
<tr>
<td>TOTPPC</td>
<td>0.412**</td>
<td>0.365**</td>
<td>0.335*</td>
<td>0.369**</td>
</tr>
</tbody>
</table>

* p < .05, ** p < .01, *** p < .001

FSIQ = intelligence quotient
MSD = motor speech disorders

The third research question of the present study was to address potential evidence of neurodegeneration in phonological working memory across ages of IWG. There was no evidence of neurodegeneration, as indicated by a decline in NRT performance with increasing age. The control group improved their performance on the NRT with increasing age at all syllable lengths, 1PPC, $\beta = .25$, p < .05, at 2PPC, $\beta = .25$, p < .001, at 3PPC, $\beta = .55$, p < .001, at 4PPC, $\beta = .45$, p < .05, and at TOTPPC, $\beta = .43$, p < .001. The galactosemia group did not improve in their NRT performance with increasing age at the single syllable length, but did improve at all other syllable lengths, 2PPC, $\beta = .66$, p < .001, at 3PPC, $\beta = .60$, p < .05, at 4PPC, $\beta = .51$, p < .05, and at TOTPPC, $\beta = .66$, p < .001.
CHAPTER IV

DISCUSSION

Deficits in phonological working memory in adults and children with galactosemia likely contribute to the high incidence of language impairment observed in this population. CWG showed poorer phonological working memory at all syllable lengths, whereas AWG showed poorer phonological working memory at the one, three, and four syllable lengths, but not at the two syllable length. This pattern differs from children with idiopathic language impairments who have poorer working memory than healthy controls at the three and four syllable lengths, but not at the one and two syllable lengths as measured by the NRT (Ellis Weismer et al., 2000). Decreased phonological working memory in IWG as compared to HC may be related to the deficit in myelin formation as a result of the inability to synthesize UDP-galactose (Ridel et al., 2005). Abnormalities in myelination in cerebral and cerebellar white matter may interfere with memory recall (Widhalm, da Cruz, & Koch, 1997).

At the single syllable length HC children in the present study averaged 91.09 (SD = 9.38) on the NRT. Children with language impairment and normal IQ averaged 90.5 (SD = 7.2), and children with borderline to low IQ had an average score of 89.3 (SD = 9.8) on the NRT (Ellis Weismer et al., 2000). CWG in the present study averaged 76.81 (SD = 15.85) and AWG averaged 84.56 (SD = 12.9) on the NRT. Dollaghan & Campbell (1998) explained that the NRT assesses an individual’s ability to transform the acoustic-phonetic sequence into its constituent phonemes and organize the articulatory input. Based on their poor performance on the NRT, IWG have difficulty organizing and maintaining the order of phonemes not only at the three and four syllable lengths, but also at the one and two syllable lengths.

One explanation for the difference in NRT performance between the IWG and HC at the
single syllable length is that IWG more frequently replaced nonwords with real words. Fifty-eight percent of participants in the CWG group and 30% of participants in the AWG group replaced nonwords with real words, compared to 20% of each control group. It is possible that IWG were experiencing programming or planning difficulties, and therefore accessed an existing motor plan for a similar familiar word, which occurred more frequently at the single syllable length, as some of these nonwords only differed from real words by one phoneme. The most common example of this pattern was replacing /voup/ with rope. The /voup/ for rope substitution accounted for 86% of real word for nonword replacements at the single syllable length in the galactosemia group, and 100% of real word for nonword replacements at the single syllable length in the control group. Lexical knowledge influences nonword repetition accuracy (Dollaghan et al., 1995) and supports repetition of more word-like nonwords, while phonological memory supports repetition of less word-like nonwords (Gathercole, 1995). Therefore, IWG may have stronger lexical knowledge than phonological memory, accounting for the replacement of more word-like nonwords by real words. Another possible explanation could be a deficiency in auditory processing. If auditory processing was deficient in IWG the participants may not have been able to distinguish minimal phonemic changes, despite normal hearing. Hearing acuity was not likely a factor in NRT performance for IWG, as most participants (32 of the 33 children and 30 of the 33 adults) had normal hearing.

Other factors may contribute to the difference in NRT performance between IWG and HC. For example, MSD and articulation disorders were prevalent in IWG, and likely affected the ability to accurately repeat nonwords. IWG diagnosed with CAS consistently scored lower on the NRT than those not diagnosed with CAS. One explanation for this finding could be articulation difficulties in those with CAS. Shriberg, Lohmeier, Campbell, Dollaghan, Green,
and Moore (2009) stated that mistakes in some nonwords may be due to misarticulations rather than inability to accurately repeat these sounds as presented. IWG with CAS had poorer articulation skills as measured by the GFTA-2. Their inconsistent articulation errors likely contributed to their performance on the NRT. Motor planning may be a contributor to poor nonword performance of children with language impairment with an MSD (Stark and Blackwell, 1997). Stark and Blackwell (1997) used nonword repetition and the imitation of isolated, repeated and sequential oral volitional movements to assess 15 children with language and articulation impairments. They found a subtle deficit in motor planning, as children with SLI were less able to perform sequences of oral movements. In the present study, no relationship between NRT performance and a diagnosis of DYS was found. DYS primarily affects motor speech execution, not motor planning (Duffy, 2005), therefore, a diagnosis of DYS did not affect NRT scores as distortions are considered correct.

IWG who scored lower on receptive language assessments also scored lower on the NRT. Dollaghan & Campbell (1998) also found that children with receptive language impairments scored lower on the NRT than children with normal language abilities. Deficits in phonological working memory contribute to low NRT scores and low receptive language skills in IWG, as the NRT is proposed to be a measure of language impairment (Coady & Evans, 2008).

Unlike previous studies of children with idiopathic language impairment, IWG with borderline to low FSIQ had decreased phonological working memory as compared to IWG with normal FSIQ scores. The NRT has been proposed to be relatively independent of IQ performance. Ellis Weismer et al. (2000) reported a significant, but modest, correlation ($r = 0.218$) between IQ and NRT scores at the three and four syllable lengths for children with language impairment. Results of the present study show a modest relationship between FSIQ
and NRT scores for one syllable length ($r = 0.282$), and stronger relationships at the three ($r = 0.314$), and four ($r = 0.394$) syllable lengths. For IWG, poor phonological working memory, even at the one and two syllable lengths, may be contributing to the individual’s lower FSIQ scores.

It should be noted that sampling bias may be present for FSIQ scores in the present study as all galactosemia participants were volunteers. Possibly, IWG who were more impacted by galactosemia were more likely to participate in the study. Interestingly, there were little differences in the mean and range of FSIQ scores for AWG and CWG. CWG had a mean FSIQ of 86, and AWG had a mean FSIQ of 88. The means and ranges in the present study are similar to those reported in previous studies (Webb et al., 2003; Waggoner et al., 1990). Participant recruitment through volunteering, rather than random selection is not necessarily a limitation of the study, as all IWG met the criteria of having been diagnosed with classic galactosemia.

The present cross-sectional study also examined the possibility of neurodegeneration with increasing age, as isolated studies have shown evidence of neurodegeneration in IWG (Jan & Wilson, 1973; Friedman, Levy, and Boustany, 1989). There was no decline in NRT performance with increasing age, but rather was a statistically significant increase in performance with increasing age at the two, three, and four syllable lengths, and at all syllable lengths in the control groups, which is indicative of improvement in phonological working memory. Manis et al. (1997) explained that although neurodegeneration may be present in a minority of IWG, it is most likely not the expected progression of the disease.

Future studies targeting specific interventions for motor planning, articulation and auditory processing are needed to determine their contributions to decrease phonological working memory in IWG. Although no previous studies have found a relationship between NRT
and motor planning, in the present study, motor planning appeared to negatively influence phonological working memory. Future studies could examine whether treatment of motor planning deficiencies in IWG with CAS will improve phonological working memory, as measured by the NRT. Treatment of phonological working memory in IWG could also be examined in future studies to see whether receptive language scores will improve as a result of therapy.
References


