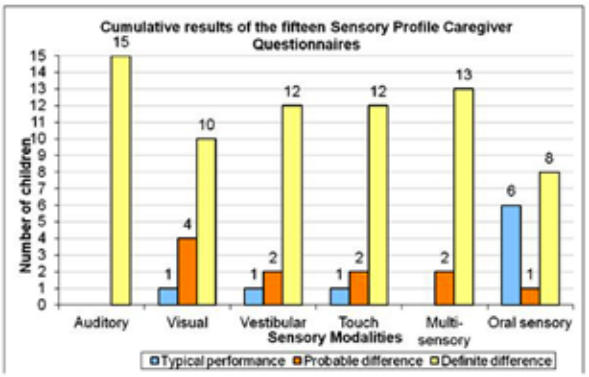
Their parents/carers completed the Sensory Profile Caregiver's Questionnaire. This is a standardised tool designed to assess children's sensory processing dysfunction in their daily functional performance.

Results There were 13 boys and 2 girls. Nine children were attending mainstream schools and six attended special schools. The assessments completed highlighted that all children experienced some form of sensory processing difficulty (Figure 1).



Abstract 243 Figure 1

Conclusion The findings support the key theme found in literature indicating that individuals with autism commonly experience sensory processing difficulties. There seem to be clear links between sensory processing difficulties and reduced functional performance during school and home activities. Further controlled studies on sensory processing in children with autism are recommended.

244 CHILD GENDER AND BIRTH ORDER INFLUENCE OUTCOMES OF AN EARLY INTERVENTION PROGRAM AT AGE 7 YEARS

doi:10.1136/archdischild-2012-302724.0244

[1,2]S Tough, [3]R Mychasiuk, [1,3]KM Benzies. *[1]Department of Paediatrics; [2]Department of Community Health Sciences; [3]Faculty of Nursing, University of Calgary, Calgary, AB, Canada*

Background and aim Early intervention programs are critical to optimize development for children in low-income families. Principles of social justice and inclusion increase the tendency to employ similar early intervention approaches for all program children. This approach fails to maximize intervention outcomes, and may benefit certain sub-groups of children more than others. The purpose of this study was to explore differences in receptive language scores in children who participated in a two-generation preschool program while controlling for child characteristics.

Method The program included centre-based care, parenting education, and family support. We assessed 62 children using the Peabody Picture Vocabulary Test III (PPVT-III) at program entry and exit, and age 7 years.

Results Repeated measures ANOVA's using child characteristics as covariates, revealed gender differences in receptive language scores at age 7 years favoring males, $F(1, 61) = 3.71$, p=0.06. Children with an older sibling exhibited significantly better receptive language scores, $F(1.61) = 4.38$, p=0.04. Ethnicity, English as a first language, time in program, and family income were unrelated to receptive language scores, p's > 0.10.

Conclusions The finding that males outperformed females is surprising because females tend to have stronger language skills than age-matched males. Younger siblings may have benefited from increased exposure to older siblings who had participated previously in the program. Results suggest that early intervention programs for children living in low-income families may benefit from alterations to program curricula that promote sex-differentiated learning strategies and focus on family dynamics.

245 SPECTRUMS AND FREQUENCIES OF SLC26A4 AND SLC26A5 GENES MUTATION AMONG PATIENTS WITH INHERITED HEARING LOSS FROM DIFFERENT REGIONS OF RUSSIA

doi:10.1136/archdischild-2012-302724.0245

L Dzhemileva, S Lobov, E Khusnutdinova. *Human Molecular Genetics, Institute of Biochemistry and Genetics, Ufa Research Center, Russian Academy of Sciences, Ufa, Russia*

Background The molecular etiology of hearing impairment in Russia has not been fully investigated. Study of *GJB2, GJÂ6, GJB3, 12SrRNA, tRNA$^{Ser(UCN)}$* and *MYO7A* genes revealed that 55% of the patients with hearing loss carried *GJB2* mutations in different regions of Russia. The *SLC26A4* and *SLC26A5* genes mutations are analyzed in this study.

Methods Two hundred and fifty unrelated deaf patients were included. The all coding exons of *SLC26A4* and first ten exons of *SLC26A5* genes were sequenced in all 250 patients, including 130 patients carrying bi- and mono-allelic recessive *GJB2* mutations, two patients carrying a known *GJB2* dominant mutation c.224G>A (p.Arg75Gln), as well as six patients with *mtDNA* (m.1555A>G, m.961insC$_{(n)}$, m.961delTinsC$_{(n)}$ and m.7444G>A) mutations.

Results Eight patients (3.2%, 8/250) with non-syndromic hearing loss were found carrying *SLC26A4* and SLC26A5 mutation and polymorphic variants. Among them, one patient with bi-allelic *SLC26A4* mutations (c.85G>C (p.Glu29Gln) and c.149T>G (p.Leu50Arg)) had EVA by CT scan. One patient with non-syndromic hearing loss was heterozygous for mutations c.919–2A>G in *SLC26A4* gene. The most common SLC26A5 gene mutation, g.-53–2A>G, accounted for 0.4% (1/250) of all *SLC26A4* mutant alleles. Two patients with non-syndromic hearing loss were heterozygous for polymorphic variant c.49548A>G (p.Gly740Ser) in *SLC26A4*, and one was heterozygous for polymorphic variant g.38190T>C in SLC26A5. The novel *SLC26A4* gene mutation g.29607delA was identified in one patient with EVA.

Conclusion Our results suggest that GJB2, SLC26A4 and SLC26A5 mutations together make up a major cause of congenital hearing loss in the different populations from Russia.

246 MATERNAL UPD2: A NEW GENETIC LOCUS FOR RUSSELL-SILVER SYNDROME

doi:10.1136/archdischild-2012-302724.0246

[1]MJ Dasouki, [2]X Tan, [3]A Patel, [2]MG Butler, Genetics. *[1]Departments of Pediatrics & Medicine; [2]Department of Psychiatry, University of Kansas Medical Center, Kansas City, KS; [3]Department of Molecular and Human Genetics, Baylor College of Medicine, Baylor University, Houston, TX, USA*

Background and aims Russell-Silver syndrome (RSS) is a genetically heterogeneous and phenotypically recognizable disorder characterized by IUGR followed by postnatal growth deficiency with head sparing, trigonocephaly, limb-length asymmetry, variable hypoglycemia, and learning disabilities. Hypomethylation of the paternal imprinting center 1 (IC1) of chromosome 11p15.5 and maternal UPD7 are identified in 35%–50% and 10% of affected individuals respectively.

Methods We studied the gDNA of a 16 month old Caucasian girl with growth failure and facial features consistent with RSS using Chromosomal Microarray Analysis (CMA) and DNA microsatellite genotyping.

Results Oligonucleotide-based CMA showed no copy number abnormality while SNP-array based CMA showed segmental Long Continuous Stretches of Homozygosity (LCSH) of 64 Mb in size involving chromosome 2 [2q11.1q13 (17.66 Mb), 2q22.1q31.1 (28.67 Mb) and 2q36.2q37.3 (17.69 Mb)]. DNA microsatellite analysis showed maternal isodisomy 2q of these regions. TIGD1 and MYEOV2 map to 2q37.1 and 2q37.3 and are predicted to be paternally expressed. However, causative imprinting of either gene was excluded since both genes map outside the smallest region of overlap between our patient and two unrelated patients with features of RSS reported by Bruno et al (J Med Genet, 2011) showing LCSH at 2q.