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).

**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 similar early intervention approaches for all program children. This study was to explore differences in receptive language scores in children living in low-income families may benefit from alterations to program curricula that promote sex-differentiated learning strategies and focus on family dynamics.

**Methods** Two hundred and fifty unrelated deaf patients were included. The all coding exons of SLC26A4 and 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, m.961delTinsC, and m.744G>A) mutations.

**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.

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**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.
Conclusions  Taken together, these results exclude possible imprinting in 2q as a cause of RSS in this child and suggest an autosomal recessive mutation which was unmasked by the segmental maternal isodisomic abnormality. Next Generation Sequence analysis of chromosome 2q regions of homozygosity identified in this child is underway and will most likely identify another novel RSS locus.

Background  NF-kB dysfunction resulting from NEMO (NF-kappaB essential modulator) mutation can lead to significant alterations in cytokine production. However, little is known about changes in the expression of downstream molecules in patients with incontinentia pigmenti (IP).

Objective  This study aims to investigate serial cytokine expressions during the first 2 years of life in young infants with IP, the period in which skin inflammation and morphological changes are most significant.

Methods  Gene analysis was performed for the two neonates with IP. Peripheral mononuclear cells were obtained shortly after birth and successively at a 6-month interval up to the age of two years. Levels of TNF-α and IL-6 were analyzed with ELISA before and after stimulating withTLR ligands.

Results  The male patient had normal NEMO allele. His cytokine level, although initially lower, had returned to a level comparable with those of controls at 12 months of age. The female infant had a mutated NEMO gene. Her baseline TNF-α level was significantly higher than those of the control subjects at birth and remained high by 6 months of age. All cytokine responses had decreased significantly by 2 years of age, the time in which all vesicular skin lesions had resolved.

Conclusion  This is the first report that demonstrates serial changes of cytokine profiles in humans with IP. This study showed that in the presence of NEMO mutation, alteration of cytokine production was remarkable during the first year of life, which may account for the prominent inflammatory changes in skin morphology.

Introduction  Glucose transporter-1 (GLUT1) deficiency syndrome (OMIM #606777) is an autosomal dominant condition resulting in reduced glucose transport into the brain. GLUT1 deficiency syndrome was first described in 1991 by De Vivo et al. The diagnostic finding is a low glucose concentration in the cerebrospinal fluid (CSF; mean 1.7 [SD 0.3mmol/L]) in the presence of normoglycaemia. GLUT1 deficiency syndrome can be confirmed by mutation analysis of the SLC2A1 gene. The spectrum is ever expanding with new mutations as also varying presentations. We present one such novel mutation.

Case report  A Caucasian male was referred to the neurology department with global developmental delay, head nods, seizures and excessive daytime sleepiness. Over the years he had many investigations including repeated MRIs and EEGs and investigations for conditions with progressive myoclonic epilepsy. At the age of 14 years the parents gave a history of food intake reducing his head nods and other seizure types. He was then investigated for possible GLUT1 deficiency.

Results  The initial investigations revealed a CSF sugar of 2.4 when the blood sugar was 5.9 (ratio of 0.4) which was low but not low enough for GLUT1 deficiency. Genetic testing revealed a mutation in the exon 5 of the SLC2A1 gene c.647T>G (p.Ile216Thr) not previously reported.

Conclusion  GLUT1 deficiency should be suspected in a child with developmental delay, epilepsy and movement disorder. Novel mutations can result in the condition. Our case is one such example for novel mutation as well as refusal of ketogenic diet from late diagnosis.