Introduction

To clarify the prevalence of UPD(16)mat in etiology-unknown patients with SRS-phenotype and phenotypic differences between UPD(16)mat and SRS.

Objectives

Methods

Results

Molecular analysis

We identified two patients (2.1%) with UPD(16)mat in 94 patients.

[Whole-exome sequencing]

• Patients 1 and 2 did not have gene mutations related to their phenotypes.

Discussion

[Phenotypical comparison between patients with UPD(16)mat in the literature and in this report and previously reported patients with SRS[1-3,5-7]]

<table>
<thead>
<tr>
<th>Phenotypic feature</th>
<th>UPD(16)mat vs. SRS (p value)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gestational age</td>
<td>0.000</td>
</tr>
<tr>
<td>Birth length</td>
<td>0.000</td>
</tr>
<tr>
<td>Birth weight</td>
<td>0.000</td>
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</tbody>
</table>

Conclusions

• Two patients (2.1%) of 94 etiology-unknown patients with SRS-phenotype had UPD(16)mat.

• We suggest considering genetic testing for UPD(16)mat in SRS-phenotype patients without known etiology.

References


Poster presented at:

2--RFC15 Growth and syndromes (to include Turner syndrome)

Takanobu Inoue

Molecular and clinical analyses of two UPD(16)mat patients detected by screening of 94 Silver-Russell syndrome patients without known etiology

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Discussion

Genetic testing for UPD(16)mat should be considered for patients with preterm birth and congenital heart diseases, even if they are not SGA.