### 28773 maternal uniparental isodisomy 14

#### Eponyms:

#### Inheritance:
- chromosomic
- genomic imprinting

#### Semeiological

#### Synthesis:
Facio-genito-ocular disorder. Mild dysmorphism, short stature, rod monochromacy inducing complete congenital achromatopsia, premature puberty. 45,XX,rob(14;14), maternal isodisomy.

#### Group Sub group Signs:

**GENITAL DISORDERS**
- genital dysfunctions
  - puberty, precocious; sexual precocity

**LABORATORY DATA**
- chromosomal disorders
  - chromosomal numerical and/or structural anomalies

**OCULAR DISORDERS**
- visus defects
  - achromatopsia, color blindness, including dyschromatopsy

**OTHERS**
- inheritance
  - inheritance, chromosomic
  - inheritance, genomic imprinting

**SKELETAL DISORDERS**
- stature, growth, modified habitus
  - stature, short, including micromelia, including short limbs

#### Super group:

#### Super aggreg.

#### Aggregations:

<table>
<thead>
<tr>
<th>Differential diagnosis</th>
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<tbody>
<tr>
<td>220</td>
<td>achromatopsia, incomplete</td>
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<tr>
<td>222</td>
<td>achromatopsia, incomplete, Smith type</td>
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<td>210</td>
<td>achromatopsia-1-2</td>
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<td>3890</td>
<td>achromatopsia-3</td>
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<tr>
<td>11900</td>
<td>congenital night blindness, stationary type 1</td>
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<td>11899</td>
<td>congenital night blindness, stationary type 3</td>
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<td>7460</td>
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<td>18830</td>
<td>Oguchi disease</td>
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#### Bibliography