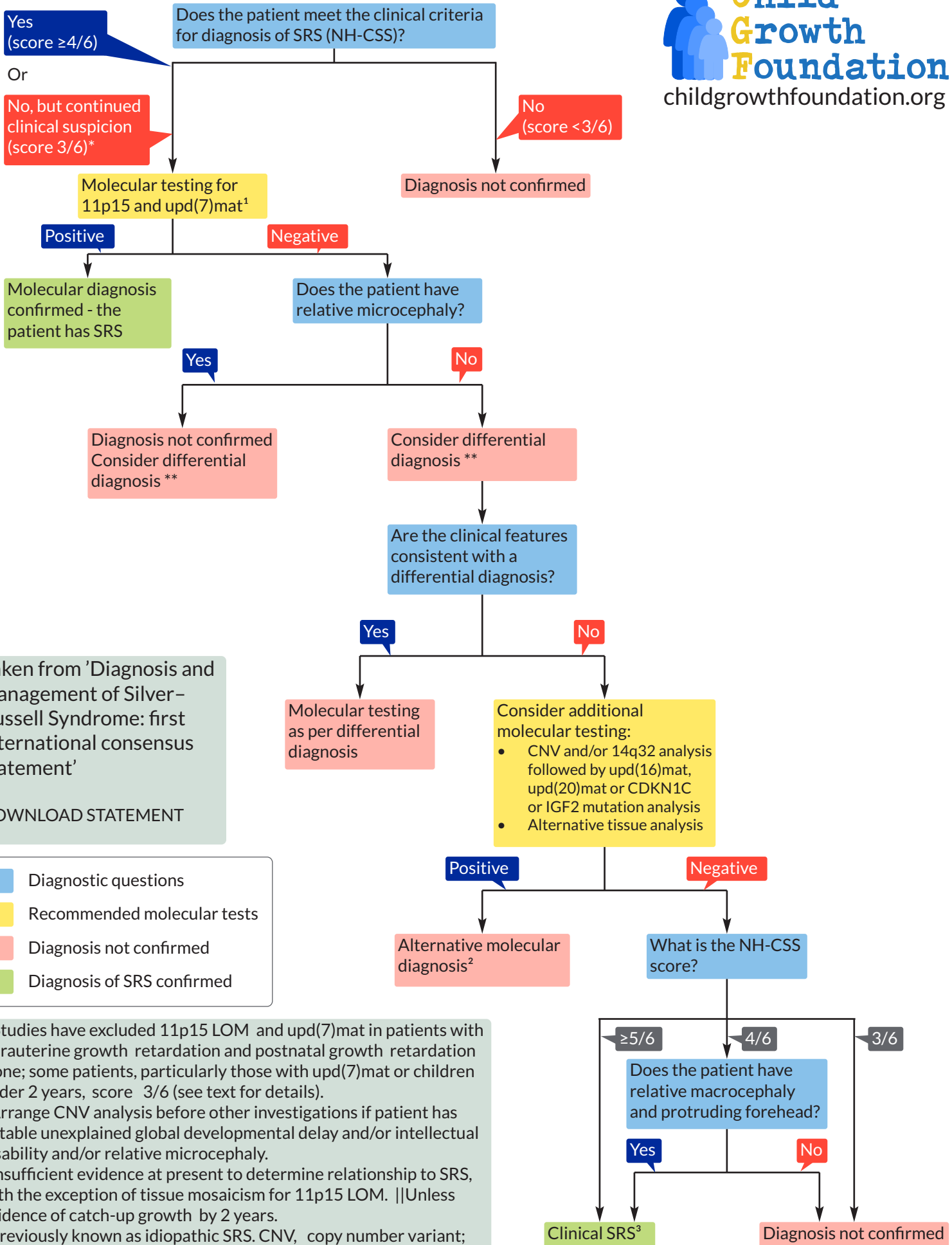


# Investigation and Diagnosis of SRS



Taken from 'Diagnosis and management of Silver-Russell Syndrome: first international consensus statement'  
 DOWNLOAD STATEMENT

- Diagnostic questions
- Recommended molecular tests
- Diagnosis not confirmed
- Diagnosis of SRS confirmed

\* Studies have excluded 11p15 LOM and upd(7)mat in patients with intrauterine growth retardation and postnatal growth retardation alone; some patients, particularly those with upd(7)mat or children under 2 years, score 3/6 (see text for details).  
<sup>1</sup> Arrange CNV analysis before other investigations if patient has notable unexplained global developmental delay and/or intellectual disability and/or relative microcephaly.  
<sup>2</sup> Insufficient evidence at present to determine relationship to SRS, with the exception of tissue mosaicism for 11p15 LOM. ||Unless evidence of catch-up growth by 2 years.  
<sup>3</sup> Previously known as idiopathic SRS. CNV, copy number variant; LOM, loss of methylation; NH-CSS, Netchine-Harbison clinical scoring system; SRS, Silver-Russell syndrome.  
 \*\* See clinical criteria for diagnosis of Silver Russell Syndrome