Reviewer's report

**Title:** Novel cAMP Binding Protein-BP (CREBBP) mutation in a girl with Rubinstein-Taybi Syndrome, GH deficiency, Arnold Chiari Malformation and Pituitary Hypoplasia.

**Version:** 4 **Date:** 28 January 2013

**Reviewer:** Anne Slavotinek

**Reviewer's report:**

Abstract:
125,000 rather than 125000
abbreviation GH should be written in full the first time the cAMP-response element binding
6 protein-BP - Óthe¹
should not be italicized
gene E1A-binding protein - Ógene¹ should not be italicized

Case presentation
2.830 - 2,830
C3546insCC - c.3546insCC
probably causes a nonsense mediated decay - probably causes nonsense mediated decay
RTS is associated with craniocervical and spinal cord complication - complications R2004X - p.Arg2004* Since also our patient presented CREBBP point mutation leading to a stop codon - ³Since our patient also had a nonsense mutation in CREBBP leading to a premature stop codon³

Conclusions
asymtomatic - ³asymptomatic²
strengthen - ³strengthen²

Fig. 1. 14 year-old - 14 years old.
Fig. 2. Arrow indicates the pituitary gland that results hypoplastic - ÓArrow indicates the pituitary gland, which is hypoplastic¹