Reviewer's report

**Title:** Shwachman-Diamond Syndrome: A complex case demonstrating the potential for misdiagnosis as Asphyxiating Thoracic Dystrophy (Jeune Syndrome)

**Version:** 1  **Date:** 24 September 2011

**Reviewer:** Ravi Savarirayan

**Reviewer's report:**

This report by Jeogh and colleagues documents a case initially diagnosed as ATD that subsequently was shown to be due to SDS. The report is well written and referenced and makes good points regarding the differing manifestations of these conditions and the need to consider the diagnosis of SDS in neonates with hypoplastic thoracic cages. The discussion points out the diagnostic confusion in the literature between ATD and SDS.

Overall a good case report making several clinically useful points.

Minor essential revision

1. Page 2. SRP includes types i-IV, please amend.

2. Figure 1. Point out the metaphyseal sclerosis present in this film, as important from diagnostic point of view.

Discretionary revision

1. Figure 3. The horizontal (flat) acetabular roofs could be pointed out in the legend.

sincerely

Professor Ravi Savarirayan

Melbourne

**Level of interest:** An article whose findings are important to those with closely related research interests

**Quality of written English:** Acceptable

**Statistical review:** No, the manuscript does not need to be seen by a statistician.

**Declaration of competing interests:**

I have no competing interests.