



**Update on the compassionate program using Translarna® (ataluren) in three patients
with Shwachman-Diamond Syndrome.**

The compassionate program based on Translarna® (ataluren) therapy in three patients with Shwachman-Diamond Syndrome (SDS) has been concluded. Two out of three patients have completed the initial 6-months period of treatment and the additional 6-months extension. One patient has completed the initial 6-months of treatment and is undergoing the 6-months extension period, which will end next summer.

Hematological assessments revealed a promising effect of Translarna®, that increased the number of platelets in all patients and improved the number of neutrophils in two out of three patients. In addition, all patients reported an increased number of B memory cells, that are generally underrepresented in SDS patients.

Bone marrow biopsies showed a remarkable increase in cellularity in all patients after 6-months of Translarna® therapy compared with the last bone marrow evaluation, performed no more than 12 months before initiation of therapy. In addition, we observed a general improvement of myeloid maturation in the absence of morphological abnormalities.

According with other clinical studies on different diseases, the drug was well tolerated by SDS patients, and no appreciable adverse events were observed.

We have discussed these results with PTC Therapeutics Incorporated. However, they communicated that the Company is not interested in further developing Translarna® for SDS. Therefore, as for the initial agreement with PTC Therapeutics, the compassionate program will end after 12 months of treatment.

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Dr Marco Cipolli, MD

