

With support from the NIH Office of Rare Diseases Research (ORDR), as well as other sources (see list below) a meeting entitled "Cockayne syndrome: Basic science and translational implications" took place in Boston, from September 12 - 15, 2009. The meeting was organized by Dr. P.J. Brooks, from the NIAAA, NIH, and Dr. Edward Neilan (Children's Hospital Boston).

The meeting was held at the Boston Hilton financial District hotel. The meeting included basic scientists and clinicians working on Cockayne syndrome from throughout the world, including Japan, France, Israel, Holland, Italy, Greece, Germany, England, Canada, Switzerland and the US. The meeting also included six families of CS patients.

The meeting began on Saturday evening, Sept. 12, with a reception and dinner, followed by brief presentations by the CS families. The first scientific session followed immediately afterward, with talks on clinical aspects of CS.

The first session on Sunday morning was on genotype phenotype relationships in CS, a particularly active area of research in recent years. This specific session was sponsored by the Ellison Foundation. The Sunday afternoon session was focused on the application of mouse models of CS. Following dinner, a short session addressed recent insights from the crystal structure of an archeal XPD protein, and the implications of these insights into diseases resulting from XPD mutations, including CS.

The scientific session on Monday morning focused in oxidative DNA damage and repair in relation to the CS phenotype, including discussion of preclinical studies with antioxidants, and possible mitochondrial defects in CS. After lunch, we had a social outing, which was an informal walk on part of the historic Freedom Trail in downtown Boston. The late afternoon session on Monday concentrated on transcriptional regulation and ubiquitination in relation to the CS.

The first morning session on Tuesday focused on translational aspects, including a presentation on the usefulness of medications intended for the treatment of Parkinson's disease in preventing some of the motor symptoms observed in CS patients. This was followed by a basic science presentation on an antioxidant compound which has shown promise in preventing some phenotypes in CS mice. The last talk in the session described the upcoming clinical trial of the compound in human CS patients.

The final session of the meeting began with an informal effort to assess the consensus of the group regarding mechanistic aspects of the CS phenotype. An overwhelming majority (>95%) agreed that the failure to repair endogenous DNA damage played at least some role in the clinical phenotype of CS. Other pathophysiologic mechanisms believed to play a role included transcriptional abnormalities, and mitochondrial dysfunction. Notably, a significant a proportion of the group (approximately 30%) felt that additional mechanisms remain to be discovered.

The remaining time was devoted to a lively "open mic" style discussion on the topic of future directions in CS research. The discussion was guided and moderated by the session chairs, and resulted in a number of insights and novel approaches to understanding CS in the future.

Based on comments from many participants, the meeting was a great success. Several scientific participants stated that this was one of the best meetings they had attended, due to the focus on CS, and, importantly, the presence of CS patients and families.

We plan to write a meeting summary paper for publication in a suitable journal.

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