## Overview

## Mitochondrial Disease Working Group: Imaging

The goal of the Mitochondrial Disorders (Mito) Imaging Common Data Elements (CDEs) Working Group (WG) was to establish a set of CDEs that would serve as a standard for mitochondrial disease clinical research within the neurological and imaging community. To do so, the Imaging WG sought to make an inclusive set of data elements to allow researchers to be highly specific in their data collection. Ideally their aim is to increase efficiency and effectiveness of clinical research studies and clinical treatment, increase data quality, and facilitate data sharing as well as help educate new clinical investigators.

The data elements created by the Imaging working group were designed to be applicable across a wide array of mitochondrial disease processes. As mentioned, the Imaging WG's goal was to establish a data form that would be all inclusive. Ideally it should serve as a reference guide on what imaging data points might be useful to collect when researching mitochondrial disorders. It is not obligatory that each data point be entered, as some studies have more focused outcomes and are only concerned with specific variables.

The WG found many gaps in existing CDEs for Imaging in mitochondrial disease. While reusing and recommending many existing CDEs for Imaging in Mitochondrial Disease the WG highlighted several variables on their Case Report Form (CRF) that are unique to though not exclusive to mitochondrial disorders. The most notable being: the deep gray nuclei, white matter tracts, and myelination pattern. An example of the discussions that arose from the creation of novel CDEs was the creation of the CDEs for myelination. Many factors went into the creation of these specific CDEs (*e.g. Should the group use a Likert scale? What existing timelines are there for myelination? What should the group use as a starting point?)* From a pediatric standpoint, it was important to the WG to put myelination in terms of age/if myelin was arrested. The final outcome was to use the permissible values (PVs) demyelination, delayed myelination, hypomyelination, and dysmyelination. The WG also made two (2) free comment myelination CDEs *distribution of abnormal myelination* and *calculated age based on myelination pattern*.

The WG created the Mitochondrial Disease Imaging CDEs as a reference guide for future research and have also created a definitions guideline to accompany their CRF which details all the elements included in their CRF as an aid for future research.

READ ME: This is a recommendations summary document of the instruments/measures/case report forms - sorted alphabetically. Details of the recommendations follow this spreadsheet in the form of information documents (e.g., Notices of Copyright) or case report forms (CRF).

**Table 1 Imaging Working Group Instrument Summary**

| Instrument / Scale / CRF Name *Name and acronym of the instrument/measure that is recommended for inclusion in the CDEs* | Domain | Subdomain | Classification (e.g., Core, Supplemental–Highly Recommended, Supplemental, Exploratory) |
|---|---|---|---|
| Brain Perfusion Magnetic Resonance Imaging | Assessments and Examinations | Imaging | Supplemental |
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