



March 2022 Rare Disease Report Updates

SETBP1 Haploinsufficiency Disorder

Reviewers: Bregie van Bon, MD, PhD, Clinical Geneticist, Radboud Medical Center

Member Organizations: [Child Neurology Foundation](#), [SETBP1 Society](#)

Central Core Disease

Reviewers: Tokunbor A. Lawal, PhD, NIH Independent Research Scholar, National Institute of Nursing Research

Member Organizations: [Muscular Dystrophy Association](#), [RYR-1 Foundation](#)

Axenfeld-Rieger Syndrome

Reviewers: Mahmoud Moustafa, MDCM Candidate, McGill University School of Medicine, and Michael Walter, PhD, Professor, University of Alberta College of Health Sciences; Faculty of Medicine and Dentistry; Department of Medical Genetics