CYP1B1 Mutations in Individuals With Primary Congenital Glaucoma and Residing in Denmark - DTU Orbit (31/03/2019)

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Primary congenital glaucoma (PCG OMIM 231300) can be caused by pathogenic sequence variations in cytochrome P450, subfamily 1, polypeptide 1 (CYP1B1). The purpose of this study was to investigate the contribution of sequence variations in CYP1B1 in a cohort of individuals with PCG residing in Denmark. The study included 37 unrelated individuals with PCG. Individuals were investigated for CYP1B1 mutations by Sanger sequencing of polymerase chain reaction products using BigDye terminators and capillary electrophoresis. A total of 12 mutations were identified and 5 of these were novel. Six were missense mutations; 4 were truncating mutations (2 nonsense and 2 frameshift); 1 was an in-frame deletion and 1 was an in-frame duplication. Mutations in CYP1B1 could fully explain the PCG phenotype in 7 individuals (18%). Five individuals were compound heterozygous or presumed compound heterozygous, 1 was homozygous and 1 was apparently homozygous. Three individuals were heterozygous for sequence variations in CYP1B1 thought to be pathogenic—one of these was p.(Tyr81Asn). Several known sequence variations with presumably no functional effect were found in the cohort. In this study, we identified 12 CYP1B1 mutations, 5 of which were novel. The frequency of CYP1B1 mutations in this cohort was comparable with other populations. We also detected an individual heterozygous for p.(Tyr81Asn) mutation, previously suggested to cause autosomal dominant primary open-angle glaucoma.

General information
State: Published
Organisations: Department of Chemistry, X-ray Crystallography
Contributors: Grønskov, K., Redó-Riveiro, A., Sandfeld, L., Zibrandtsen, N., Harris, P., Bach-Holm, D., Türmer, Z.
Pages: 926–930
Publication date: 2016
Peer-reviewed: Yes

Publication information
Journal: Journal of Glaucoma
Volume: 25
Issue number: 12
ISSN (Print): 1057-0829
Ratings:
BFI (2019): BFI-level 1
Web of Science (2019): Indexed yes
BFI (2018): BFI-level 1
Web of Science (2018): Indexed yes
BFI (2017): BFI-level 1
Scopus rating (2017): CiteScore 1.82 SJR 1.08 SNIP 1.061
Web of Science (2017): Impact factor 1.742
Web of Science (2017): Indexed yes
BFI (2016): BFI-level 1
Scopus rating (2016): CiteScore 2.06 SJR 1.176 SNIP 1.314
Web of Science (2016): Impact factor 2.263
Web of Science (2016): Indexed yes
BFI (2015): BFI-level 1
Scopus rating (2015): CiteScore 1.98 SJR 1.417 SNIP 1.216
Web of Science (2015): Impact factor 2.102
BFI (2014): BFI-level 1
Scopus rating (2014): CiteScore 1.82 SJR 1.254 SNIP 1.263
Web of Science (2014): Impact factor 2.106
BFI (2013): BFI-level 1
Scopus rating (2013): CiteScore 2.01 SJR 1.498 SNIP 1.295
Web of Science (2013): Impact factor 2.427
BFI (2012): BFI-level 1
Scopus rating (2012): CiteScore 1.85 SJR 1.725 SNIP 1.35
Web of Science (2012): Impact factor 1.865
BFI (2011): BFI-level 1
Scopus rating (2011): CiteScore 2.1 SJR 1.791 SNIP 1.355
Web of Science (2011): Impact factor 1.776