ACCSCIENCE PUBLISHING

Gene & Protein in Disease

ORIGINAL RESEARCH ARTICLE

Genetic analysis of *GJA3* and *GJA8* mutations in cataract patients from the Jammu region, North India

Supplementary Files

Table S1. Mutations in the *GJA3* gene

Gene	Nucleotide	Mutation	Cataract type	Family origin	References
GJA3	c. 7 G>T	p.D3Y	Zonular pulverulent cataract	Hispanic	1
GJA3	c. 32T>C	p.L11S	Ant-egg, lamellar cataract	Danish	2
GJA3	C. 82G>A	p.V28M	Variable total cataract	Indian	3
GJA3	c. 96C>A	p.F32L	Nuclear pulverulent	Chinese	4
GJA3	c. 98G>T	p.R33L	Embryonal nuclear granular	Indian	5
GJA3	c. 130G>A	p.V44M	Nuclear cataract	Chinese	6
GJA3	c. 134G>C	p.W45S	Nuclear progressive cataract	Chinese	7
GJA3	c. 139G>A	p.D47N	Nuclear central cataract	Chinese	8
GJA3	c. 176C>T	p.P59L	Nuclear punctate	American	9
GJA3	c. 226G>C	p.R76G	Total	Indian	3
GJA3	c. 227G>A	p.R76H	Zonular pulverulent	Australian	10
GJA3	c. 260C>T	p.T87M	Pearl box nuclear cataract	Indian	5
GJA3	c. 560C->T	p.P187L	Zonular pulverulent	Caucasian	11
GJA3	c. 559C>T	p.P187S	Nuclear pulverulent	Chinese	12
GJA3	c. 563A>C	p.N188T	Nuclear pulverulent	Chinese	13
GJA3	c. 563A>T	p.N188I	Zonular pulverulent	Chinese	14
GJA3	c. 5G>A	p.G2D	Nuclear pulverulent and posterior polar	Chinese	15
GJA3	c. 56C>T	p.T19M	Posterior polar	Indian	16
GJA3	c. 134G>C	p.W45S	Nuclear	Chinese	7
GJA3	c. 616T>A	p.F206I	Nuclear	Chinese	17
GJA3	c. 139G>A	p.D47N	Nuclear	Chinese	18
GJA3	c. 134G→C	p. W 45S	Nuclear cataracts	Chinese	19
GJA3	c. 130G>A	p.V44M	Autosomal dominant cataract	Caucasian	9

Table S2. Mutations in the *GJA8* gene

Gene	Mutation	Cataract type	Family origin	References
GJA8	c. 68G>C	Progressive dense nuclear	Iranian	20
GJA8	c. 131T>A	Cataract and microcornea	Indian	21
GJA8	c. 134G>C	Jellyfish-like cataract and microcornea	Indian	22
GJA8	c. 593G>A	Posterior subcapsular cataract	Indian	21
GJA8	c. 565C>T	Nuclear cataract and microcornea	Danish	2
GJA8	c. 905T>C	Zonular cataract	India	23
GJA8	c. 670insA	Total cataract and nystagmus	Indian	24
GJA8	c. 741T>G	Zonular pulverulent cataract	Russian	25
GJA8	ins776G	Triangular nuclear cataract	German	26
GJA8	c. 827C>T	Pulverate nuclear cataract	Chinese	27
GJA8	c. 142G>A	Zonular nuclear pulverulent	Pakistani	28
GJA8	c. 235G>C	Full moon like with Y-sutural opacities	Indian	29
GJA8	c. 262C>T	Zonular pulverulent	English	30
GJA8	c. 759C>T	Lamellar cataract/total cataract	North Indian	Present study

Table S3. Reporting checklist for the case–control study

Features	Item	Reporting item	Status
Title and abstract			
Title	#1a	Indicate the study's design with a commonly used term in the title or the abstract	✓
Abstract	#1b	Provide in the abstract an informative and balanced summary of what was done and what was found	✓
Introduction			
Background/rationale	#2	Explain the scientific background and rationale for the investigation being reported	✓
Objectives	#3	State-specific objectives, including any pre-specified hypotheses	✓
Methods			
Study design	#4	Present key elements of the study design early in the paper	✓
Setting	#5	Describe the setting, locations, and relevant dates, including periods of recruitment, exposure, follow-up, and data collection	✓
Eligibility criteria	#6a	Give the eligibility criteria, and the sources and methods of case ascertainment and control selection. Give the rationale for the choice of cases and controls. For matched studies, give the matching criteria and the number of controls per case	✓
Eligibility criteria	#6b	For matched studies, give the matching criteria and the number of controls per case	✓
	#7	Clearly define all outcomes, exposures, predictors, potential confounders, and effect modifiers. Give diagnostic criteria, if applicable	✓
Data sources/ measurement	#8	For each variable of interest, give sources of data and details of methods of assessment (measurement). Describe the comparability of assessment methods if there is more than one group. Give information separately for cases and controls	✓
Bias	#9	Describe any efforts to address potential sources of bias	NA
Study size	#10	Explain how the study size was arrived at	X
Quantitative variables	#11	Explain how quantitative variables were handled in the analyses. If applicable, describe which groupings were chosen and why	NA
Statistical methods	#12a	Describe all statistical methods, including those used to control for confounding	✓
Statistical methods	#12b	Describe any methods used to examine subgroups and interactions	NA
Statistical methods	#12c	Explain how the missing data were addressed	NA
Statistical methods	#12d	If applicable, explain how the matching of cases and controls was addressed	✓

(Cont'd...)

Table S3. ***(Continued)***

Features	Item	Reporting item	Status
Statistical methods	#12e	Describe any sensitivity analyses	NA
Results			
Participants	#13a	Report numbers of individuals at each stage of study—e.g., numbers potentially eligible, examined for eligibility, confirmed eligible, included in the study, completing follow-up, and analysed. Give information separately for cases and controls	NA
Participants	#13b	Give reasons for non-participation at each stage	NA
Participants	#13c	Consider the use of a flow diagram	NA
Descriptive data	#14a	Give characteristics of study participants (e.g., demographic, clinical, social) and information on exposures and potential confounders. Give information separately for cases and controls	
Descriptive data	#14b	Indicate the number of participants with missing data for each variable of interest	NA
Outcome data	#15	Report numbers in each exposure category, or summary measures of exposure. Give information separately for cases and controls	✓
Main results	#16a	Give unadjusted estimates and, if applicable, confounder-adjusted estimates and their precision (e.g., 95% confidence interval). Make clear which confounders were adjusted for and why they were included	X
Main results	#16b	Report category boundaries when continuous variables were categorized	NA
Main results	#16c	If relevant, consider translating estimates of relative risk into absolute risk for a meaningful time period	NA
Other analyses	#17	Report other analyses done—e.g., analyses of subgroups and interactions, and sensitivity analyses	NA
Discussion			
Key results	#18	Summarize key results with reference to study objectives	✓
Limitations	#19	Discuss limitations of the study, taking into account sources of potential bias or imprecision. Discuss both the direction and the magnitude of any potential bias	✓
Interpretation	#20	Give a cautious overall interpretation considering objectives, limitations, multiplicity of analyses, results from similar studies, and other relevant evidence	NA
Generalizability	#21	Discuss the generalizability (external validity) of the study results	NA
Other information			
Funding	#22	Give the source of funding and the role of the funders for the present study and, if applicable, for the original study on which the present article is based	✓

Abbreviation: NA: Not available.

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