Data in Brief 10 (2017) 406–412

Contents lists available at ScienceDirect

Data in Brief

journal homepage: www.elsevier.com/locate/dib

Data Article

Data on the 21-Hydroxylase deficient CAH patients and the identification of known/novel mutations in CYP21A2 gene

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ARTICLE INFO

Article history:
Received 24 November 2016
Received in revised form
3 December 2016
Accepted 6 December 2016
Available online 15 December 2016

Keywords:
CYP21A2 gene
Salt wasting
Simple virilizing
Non classical
Known mutations
Novel mutations

ABSTRACT

This article presents the dataset regarding spectrum of mutations in 21-Hydroxylase deficient CAH patients as described in "The spectrum of CYP21A2 mutations in Congenital Adrenal Hyperplasia in an Indian cohort" (R. Khajuria, R. Walia, A. Bhansali, R. Prasad, 2017) [1]. This dataset features about the CAH patients in the cohort, their classification into subtypes and finally screening the exon–intron boundaries of 21-Hydroxylase gene (CYP21A2) to detect common mutations, novel mutations along polymorphisms in the CYP21A2 gene. The specified large set of primers and the parameters for the mutation detection allow the identification and molecular characterization of CYP21A2 gene in the CAH patients.

Specifications Table

Subject area	Biology
More specific subject area	Endocrinology, Molecular biology

DOI of original article: http://dx.doi.org/10.1016/j.cca.2016.11.037
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http://dx.doi.org/10.1016/j.dib.2016.12.013

Type of data	Text, Table, Graph, Figure
How data was acquired	Scatter diagram for 17-α-OHP, Primer sequences were checked using BLAST search
Data format	Analyzed
Experimental factors	DNA isolated from the blood of CAH patients
Experimental features	ELISA, Polymerase chain reaction
Data source location	Chandigarh, India
Data accessibility	Data is with this article and available at genbank via accession numbers: NCBI accession number-KF812549, NCBI accession number- KF534754, NCBI accession number- KF692099, NCBI accession number- KF447378

Value of the data

- The data provides the information about female to male ratio in the 21-Hydroxylase deficient CAH patients which could be compared to other studies.
- The data supports that level of 17-α-OHP in classical CAH patients is higher than non classical CAH patients as mentioned by other researchers and clinicians.
- The sequence of the primers mentioned would help other researchers to identify common and novel mutations in CYP21A2 gene.

1. Data

Congenital Adrenal Hyperplasia is an autosomal recessive disorder mainly caused by defects in 21-Hydroxylase gene (CYP21A2) which codes for 21-Hydroxylase enzyme [2]. Figs. 1–3 and Table 1 indicate ratio of patients (males and females) in classical (SW, SV) and non classical CAH and the associated level of 17-α-OHP which is the substrate of 21-Hydroxylase enzyme. The major disease-causing mutations in CYP21A2 (functional gene) are transferred from CYP21A1P (pseudogene) due to unequal crossing over during meiosis or apparent gene conversion events [3], macro or micro-conversion events [4]. Tables 1–5 elucidate the age of the CAH patients in the classical and non-classical CAH, the primer sequences which were used for detection of the common mutations,

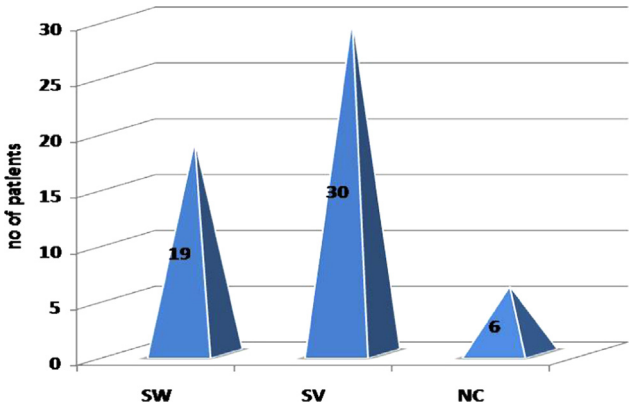

Fig. 1. Number of patients in each sub-type of CAH.

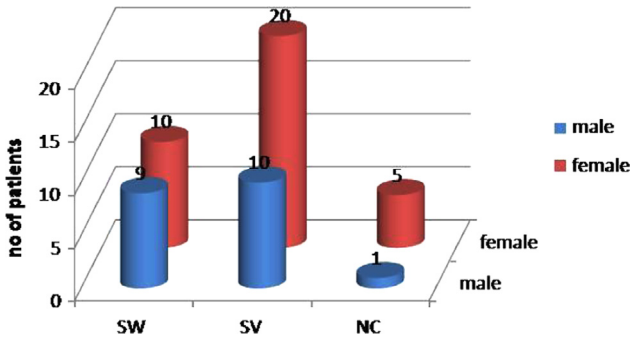

Fig. 2. Number of patients in each sub-type of CAH along the male to female ratio in the three sub-types of CAH.

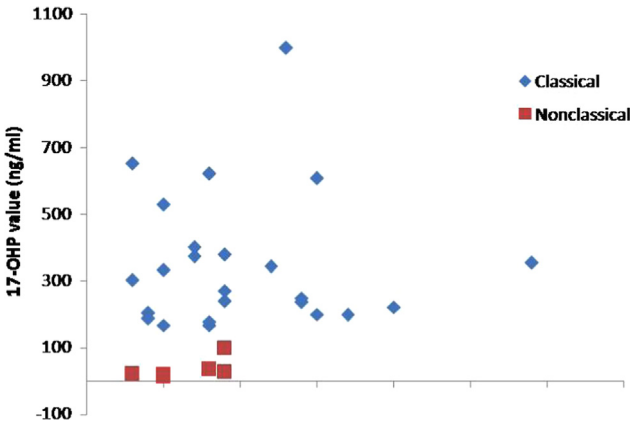

Fig. 3. Scatter diagram representing the level of 17-α-OHP (ng/ml) in different categories of CAH (classical and non-classical). Levels of 17-α-OHP are higher in classical form of CAH as compared to non-classical form of CAH.

Table 1
Range of age in CAH patients.

Category of CAH	Minimum age (years)	Maximum age (years)
Salt wasting	1 month	19
Simple virilizing	3.5	55
Non classical	17	24

polymorphisms and novel mutations in the CYP21A2 gene. The novel mutations were detected at the frequency of 3%–5% when large cohorts were investigated [5] (Table 6).

2. Experimental design

2.1. Sample collection

The patients were categorized into 3 types viz., Salt Wasting (SW), Simple Virilizing (SV) and non classical (NC). CAH patients had varied age groups among the 3 types.

Table 2
Oligonucleotides (primers) used to amplify the CYP21A2 gene including exon-intron boundaries. Column 1 is primer code, column 2 is primer sequence, column 3 is PCR product size (bp), Column 4 is mutation detected and column 5 is annealing temperatures(°C).

1	2	3	4	5
P 1	5′-TGC ATT TCC CTT CCT TGC TTC-3′	952	F1	63.2
P 2	5′-GCA GGG AGT AGT CTC CCA AGG-3′[a]			
P 3	5′-CCT TGG GAG ACT ACT CCC TGC-3′	320	I172N E6 cluster	58.4
P 4	5′-AGG GGT TCG TAC GGG AGC AAT A-3′[a]	2070	F2	64.2
P 5	5′-CTG AGG TGC CAC TTA TAG CTC-3′[a]			
P 6	5′-AAG CTC CGG AGC CTC CAC CTC G-5′	148	P30L	51.5
P 7	5′-AGA TCA GCC TCT CAC CTT GC-3′[a]			
P 8	5′-TGG GGC ATC CCC AAT CCA GGT CCC-3′	156	i2g	62.0
P 9	5′-ACC AGC TTG TCT GCA GGA GGAT-3′[a]			
P 10	5′-TCT CCG AAG GTG AGG TAA CA T-3′[a]	320	I172N	58.4
P 11b	5′-AGC TGC ATC TCC ACG ATG GA-3′[a]	696	E6 cluster N allele	60.6
P 11a	5′-TCA GCT GCT TCT CCT CGT TGT GG-3′[a]	696	E6 cluster M allele	60.2
P 12	5′-GAT CAC ATC GTG GAG ATG CAG CTG-3′	781	V281L	71.0
P 13	5′TGG GCC GTG TGG TGC GGT GGG GCA A-3′[a]		Q318X	
P 14	5′CCA GAT TCA GCA GCG ACT G-3′	162	R356W	67.0
P 15	5′-TGG GGC AAG GCT AAG GGC ACA AC C-3′[a]			

[a] underlined primers are antisense primers.

Table 3
List of the mutations, PCR product size, the restriction enzyme used and the fragment size obtained after digestion for the detection of the common mutations.

Mutation	PCR product	Restriction enzyme	Fragments produced after digestion if mutation present	Fragments produced after digestion if mutation absent
P30L	148 bp	Bsh12361	148 bp	126 bp & 22 bp
I172N	320 bp	Nde 1	297 bp & 28 bp	320 bp
V281L	781 bp	Apa L1	686 bp & 95 bp	375 bp,311 bp & 95 bp
i2g	156 bp	Sau3A1	133 bp & 23 bp	156 bp
Q318X	781 bp	Pst 1	457 bp,204 bp & 120 bp	299 bp,204 bp,158 bp & 120 bp
R356W	163 bp	Eco521	162 bp	136 bp
Gene Deletion	210 bp	Taq 1	187 bp	210 bp & 187 bp

2.2. 17-α-OHP measurement

17-α-OHP was measured in the serum samples of the CAH patients by enzyme-linked immunesorbent assay (ELISA), based on the principle of competitive binding.

2.3. Identification of common mutations, polymorphisms and novel mutations

DNA was isolated by the standard protocols [6]. The common mutations, polymorphisms and the novel mutations were detected the 110 alleles [1]. Various mutations were present at different

Table 4
List of the mutations, optimum temperature, time duration required for the detection of the known mutations.

Mutation	Restriction enzyme	Temp.	Duration of incubation
P30L	Bsh12361	37 °C	4 h
I172N	Nde 1	37 °C	4 h
V281L	Apa L1	37 °C	4 h
i2g	Sau3A1	37 °C	2 h
Q318X	Pst 1	37 °C	4 h
R356W	Eco521	37 °C	4 h
Gene deletion	Taq 1	65 °C	4 h

Table 5
Mutations or sequence variations, primers used for PCR and restriction enzymes used in detection of normal and mutant alleles.

Poly-morphim	Primer	PCR product	Enzyme	Fragment Size (bp)	
				Normal allele	Mutant allele
S268T	7-F TGCAGGAGAGCCTCGTGGCAGG 7-R ACGCACCTCAGGGTGGTGAAG	212 bp	Nco 1	–	–
D183E	5-F GGAGACAAGATCAAGGTGCCT 5-R CCAGGTCCTCACCCTGAGA	217 bp	–	212	146 and 66

Table 6
Oligonucleotides primers used for amplification of CYP21A2 gene exons.

Exons	Primer Sequence 5′–3′	PCR product (bp)	Annealing temperature (°C)
Forward 1	AGCGGATCCCCCGGTGGCCTC	216	63.0
Reverse 1	CCGTGGCCCAGCCTGCAGATG		
Forward 2	AGCTCTGAGGACTGATCTTGA	208	61.8
Reverse 2	CCGTGGCCCAGCCTGCAGATG		
Forward 3	AGCTCTGAGGACTGATCTTGA	226	66.4
Reverse 3	AGCAGCAGTTGGAGCCAGGTT		
Forward 4	GTACGATAGCACCTTCCTGTT	207	61.8
Reverse 4	GCTGAGTCTCCAACTCTGGTT		
Forward 5	TTGGGGTTCGCCCTGCCCGTA	217	68.6
Reverse 5	CAAAGCTTCATCACCCCCTCC		
Forward 6	AGGAGGGAGTTGACTTGGTGT	193	63.4
Reverse 6	CTGTTCCCATGTCCACAGTGC		
Forward 7	TGGGACAGAGGAAATATGCCA	212	65.5
Reverse 7	CCTTTACZCACCTCTCTCATG		
Forward 8	GGCTCCTATGTCACCTTGATG	227	62.1
Reverse 8	CAACCTCCATCCAGTGCCTAG		
Forward 9	GGTCAGCATCTGGACCCCAGG	212	66.9
Reverse 9	AGGTTGCAGTTCACTAGGCTG		
Forward 10	AGGTGCTAACCTGGATAACTG	303	59.8
Reverse 10	CACATACTGCATGTGAGAGTC		

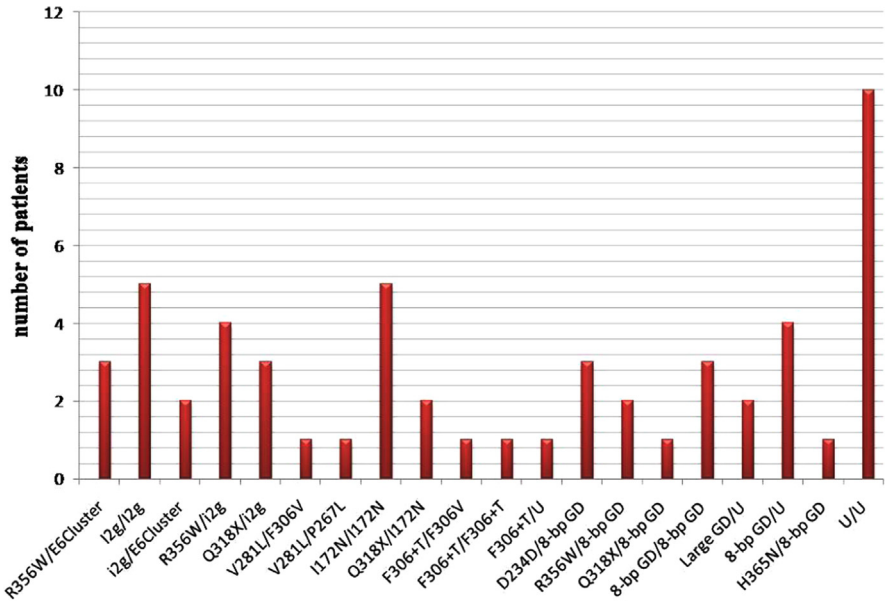

Fig. 4. Type of genotype and their abundance in Indian CAH patients.

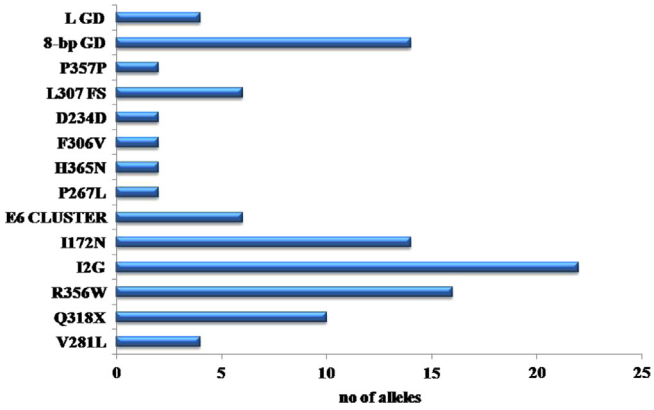

Fig. 5. Bar diagram representing various mutations identified and the corresponding number of mutated alleles affected with each mutation.

frequencies in our population [1]. The genotype of the patients and the affected no of alleles were detected in the present study (Figs. 4, 5). The prevalence of common mutations in 3 sub-types of CAH were also studied in present study (Fig. 6).

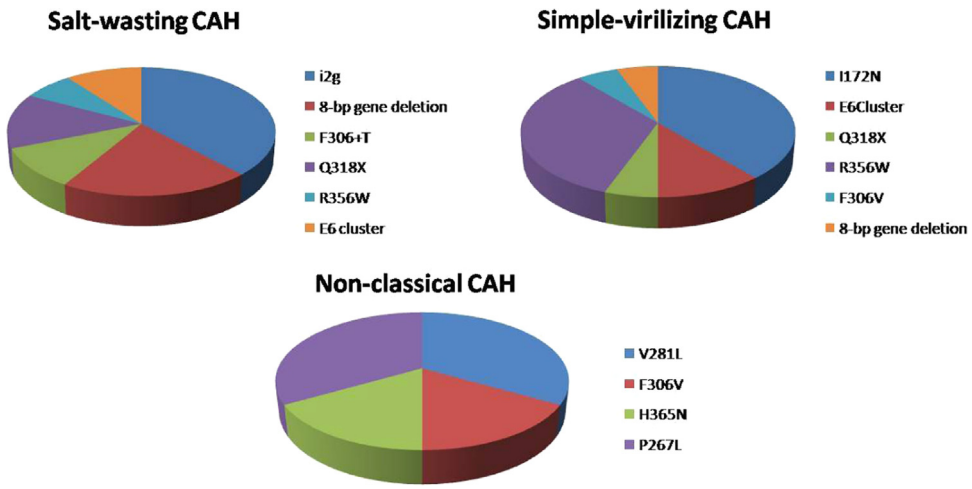

Fig. 6. Mutations prevailing in different forms of CAH- Salt-Wasting, Simple-Virilizing and Non-Classical respectively in our population.

Acknowledgments

We express our sincere gratitude to Science & Engineering Research Board (SERB), New Delhi, India for funding the project (Sanction number: SR/SO/HS/0045/2010) and Indian Council of Medical Research, New Delhi for ICMR-JRF and SRF (Reference number: 3/1/3/JRF-2011/HRD-3). We are also thankful to all the patients included in this study.

Transparency document. Supporting Material

Transparency data associated with this article can be found in the online version at http://dx.doi.org/10.1016/j.dib.2016.12.013.

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