Rare Disease Profiling

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Family History

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Listed of Disease/Condition in this Report

--- Immunodeficiency due to a late component of complement deficiency
--- Normosmic congenital hypogonadotropic hypogonadism
--- Fleck corneal dystrophy
--- Hereditary breast cancer
--- Adams-Oliver syndrome
--- Amyotrophic lateral sclerosis
--- Ellis Van Creveld syndrome
--- Osteogenesis imperfecta type 3
--- Localized junctional epidermolysis bullosa, non-Herlitz type
--- Porencephaly
--- Spondyloepimetaphyseal dysplasia, aggrecan type
--- Glycogen storage disease due to acid maltase deficiency
--- Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome

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Report Disclaimer

Lorem ipsum dolor sit amet, consectetur adipiscing elit. Mauris vehicula libero at sollicitudin dignissim. Nam aliquam arcu ut risus tempus facilisis. Etiam congue tempus tellus. Nulla eu enim a mi sagittis sagittis nec bibendum quam. Proin eget pulvinar tortor. Fusce nibh purus, tempus vitae hendrerit id, pretium sit amet eros. Cras eget dui justo. Ut interdum pulvinar facilisis. Quisque luctus efficitur nisi eu imperdiet. Aliquam fermentum enim non elit aliquet eleifend. Sed nunc sem, tempor in venenatis in, volutpat non magna. Suspendisse sodales, purus nec imperdiet molestie, tortor magna tristique nisi, id pulvinar eros lorem quis lacus. Cras eu augue lorem. Pellentesque at turpis eget nunc sollicitudin semper sed at dolor. Nulla tristique maximus tellus at finibus. Vestibulum eu erat sit amet enim tristique condimentum at sed erat. Ut massa quam, euismod non dictum ut, vehicula quis quam. Nullam id lorem maximus, imperdiet neque eget, tristique mauris. Vivamus accumsan mollis lacus et scelerisque. Suspendisse elementum ex sed dui interdum gravida. Fusce vel risus orci. Fusce efficitur, odio id luctus placerat, metus orci sollicitudin mauris, a sagittis urna nulla facilisis nibh. Maecenas luctus semper libero, quis lobortis ligula placerat eu. Sed commodo orci lectus, a luctus tellus lobortis a. Duis sit amet tempus metus. Proin semper ac leo et aliquet. Vestibulum porttitor et orci ut commodo. Praesent sed bibendum arcu. Integer rhoncus magna interdum, rutrum augue vel, rutrum turpis. Nulla ut nisi ornare, viverra nisi ut, egestas ipsum. Nam porttitor risus id lorem elementum porttitor. Curabitur sollicitudin metus non euismod ullamcorper. Orci varius natoque penatibus et magnis dis parturient montes, nascetur ridiculus mus. Phasellus neque nulla, congue sed consequat ac, elementum id nulla. Mauris nec porttitor purus. Mauris dapibus eget magna et tempus. Morbi tincidunt, sapien vel mattis blandit, elit nulla sodales mi, quis semper ante eros vitae nunc. Phasellus euismod diam id urna pulvinar condimentum id non lacus. Ut pretium facilisis tortor. Sed eget orci fringilla tortor faucibus luctus euismod quis velit. Suspendisse lorem diam, pellentesque sed sollicitudin at, semper vel leo. Proin id luctus risus. Donec accumsan odio quis nunc ultricies, sed tincidunt metus porta. Vestibulum interdum dui lectus, in placerat nisi tempor quis. Maecenas at erat in nunc tincidunt faucibus eget vitae mauris. Integer ultricies tristique dui non convallis. Curabitur tempor vehicula lacus, sit amet fringilla nisl efficitur a. Fusce vitae faucibus risus. Curabitur ut nulla eget sapien fermentum eleifend. Proin ante magna, pharetra ac tempor ac, lobortis in erat. Nam ipsum tortor, venenatis semper ligula ut, pharetra finibus massa. Fusce euismod vulputate tellus vitae pellentesque. Fusce sit amet tincidunt mi. Aliquam erat volutpat. Pellentesque habitant morbi tristique senectus et netus et malesuada fames ac turpis egestas. Mauris vel semper metus. Sed tortor metus, aliquam non eleifend at, convallis id ligula. Nam fringilla, est non mollis semper, turpis arcu iaculis neque, in sagittis libero leo ut sapien. Maecenas maximus pharetra magna non molestie. Mauris euismod fermentum varius. Donec pulvinar sit amet lorem at maximus. Donec vestibulum nibh eu varius tincidunt.

Name:	: John Doe	Patient ID	: ID20240320
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Date of Birth	: 01/01/1990		

Immunodeficiency due to a late component of complement deficiency

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr1	56854927	T	C	24.1	QUAL30	Uncertain significance	Unknown
chr1	56875054	C	A	31.1	PASS	Benign	Benign
chr1	56956811	C	T	58.6	PASS	Benign	Unknown
chr5	39364452	G	A	21.4	QUAL30	Benign	Benign
chr5	40955459	G	C	67.6	PASS	Benign	Unknown
chr5	41158761	G	A	49.8	PASS	Benign	Unknown
chr5	41161851	T	C	16.9	QUAL30	Likely benign	Unknown
chr9	120963693	C	T	20.0	QUAL30	Benign	Unknown
chr9	121021656	T	C	15.4	QUAL30	Benign	Benign
chr9	121037940	C	T	11.7	QUAL30	Benign	Unknown

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr1	56854927	T	C	24.1	QUAL30	.	0.000798722
chr1	56875054	C	A	31.1	PASS	0.3259	0.34365
chr1	56956811	C	T	58.6	PASS	0.9785	0.986422
chr5	39364452	G	A	21.4	QUAL30	0.3422	0.286142
chr5	40955459	G	C	67.6	PASS	0.5283	0.48123
chr5	41158761	G	A	49.8	PASS	0.9996	0.998203
chr5	41161851	T	C	16.9	QUAL30	0.0008	0.00419329
chr9	120963693	C	T	20.0	QUAL30	0.0784	0.106629
chr9	121021656	T	C	15.4	QUAL30	0.2403	0.241214
chr9	121037940	C	T	11.7	QUAL30	0.1334	0.146565

Name:	: John Doe	Patient ID	: ID20240320
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Immunodeficiency due to a late component of complement deficiency

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr1	56854927	T	C	non synonymous SNV	-
chr1	56875054	C	A	non synonymous SNV	-
chr1	56956811	C	T	non synonymous SNV	-
chr5	39364452	G	A	non synonymous SNV	-
chr5	40955459	G	C	non synonymous SNV	-
chr5	41158761	G	A	synonymous SNV	-
chr5	41161851	T	C	non synonymous SNV	-
chr9	120963693	C	T	synonymous SNV	-
chr9	121021656	T	C	synonymous SNV	-
chr9	121037940	C	T	non synonymous SNV	-

Name:	: John Doe	Patient ID	: ID20240320
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Immunodeficiency due to a late component of complement deficiency

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr1	56854927	T	C	-	-
chr1	56875054	C	A	-	-
chr1	56956811	C	T	-	-
chr5	39364452	G	A	-	-
chr5	40955459	G	C	-	-
chr5	41158761	G	A	-	-
chr5	41161851	T	C	-	-
chr9	120963693	C	T	-	-
chr9	121021656	T	C	-	-
chr9	121037940	C	T	-	-

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
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Normosmic congenital hypogonadotropic hypogonadism

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr2	128318387	T	C	37.8	PASS	Benign	Unknown
chr4	67744591	C	T	20.7	QUAL30	Uncertain significance	Conflicting
chr8	25423284	C	G	44.7	PASS	Benign	Benign
chr10	120889970	A	G	13.1	QUAL30	Benign	Unknown
chr10	120904073	G	A	29.0	QUAL30	Benign	Unknown
chr12	89351700	C	A	24.3	QUAL30	Benign	Benign
chr19	920642	T	A	25.9	QUAL30	Benign	Benign
chr20	5302610	C	G	18.8	QUAL30	Benign	Benign
chrX	8535600	G	A	66.2	PASS	Benign	Benign
chrX	8536792	C	T	63.3	PASS	Benign	Benign

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr2	128318387	T	C	37.8	PASS	0.6869	0.723442
chr4	67744591	C	T	20.7	QUAL30	6.462e-05	0.00139776
chr8	25423284	C	G	44.7	PASS	0.2175	0.224241
chr10	120889970	A	G	13.1	QUAL30	0.2203	0.197085
chr10	120904073	G	A	29.0	QUAL30	0.7080	0.676518
chr12	89351700	C	A	24.3	QUAL30	0.4826	0.466254
chr19	920642	T	A	25.9	QUAL30	0.7992	0.783946
chr20	5302610	C	G	18.8	QUAL30	0.7294	0.723243
chrX	8535600	G	A	66.2	PASS	0.5496	0.481854
chrX	8536792	C	T	63.3	PASS	0.5582	0.488212

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Normosmic congenital hypogonadotropic hypogonadism

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr2	128318387	T	C	synonymous SNV	Autosomal dominant, Autosomal recessive, X-linked recessive, Multigenic/multifactorial
chr4	67744591	C	T	non synonymous SNV	Autosomal dominant, Autosomal recessive, X-linked recessive, Multigenic/multifactorial
chr8	25423284	C	G	non synonymous SNV	Autosomal dominant, Autosomal recessive, X-linked recessive, Multigenic/multifactorial
chr10	120889970	A	G	synonymous SNV	Autosomal dominant, Autosomal recessive, X-linked recessive, Multigenic/multifactorial
chr10	120904073	G	A	synonymous SNV	Autosomal dominant, Autosomal recessive, X-linked recessive, Multigenic/multifactorial
chr12	89351700	C	A	non synonymous SNV	Autosomal dominant, Autosomal recessive, X-linked recessive, Multigenic/multifactorial
chr19	920642	T	A	non synonymous SNV	Autosomal dominant, Autosomal recessive, X-linked recessive, Multigenic/multifactorial
chr20	5302610	C	G	synonymous SNV	Autosomal dominant, Autosomal recessive, X-linked recessive, Multigenic/multifactorial
chrX	8535600	G	A	synonymous SNV	Autosomal dominant, Autosomal recessive, X-linked recessive, Multigenic/multifactorial
chrX	8536792	C	T	non synonymous SNV	Autosomal dominant, Autosomal recessive, X-linked recessive, Multigenic/multifactorial

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Normosmic congenital hypogonadotropic hypogonadism

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr2	128318387	T	C	Infancy, Neonatal	-
chr4	67744591	C	T	Infancy, Neonatal	-
chr8	25423284	C	G	Infancy, Neonatal	-
chr10	120889970	A	G	Infancy, Neonatal	-
chr10	120904073	G	A	Infancy, Neonatal	-
chr12	89351700	C	A	Infancy, Neonatal	-
chr19	920642	T	A	Infancy, Neonatal	-
chr20	5302610	C	G	Infancy, Neonatal	-
chrX	8535600	G	A	Infancy, Neonatal	-
chrX	8536792	C	T	Infancy, Neonatal	-

Name:	: John Doe	Patient ID	: ID20240320
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Date of Birth	: 01/01/1990		

Fleck corneal dystrophy

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr2	208320256	G	A	50.6	PASS	Benign	Benign
chr2	208320275	C	T	53.9	PASS	Benign	Benign
chr2	208325606	T	C	67.7	PASS	Benign	Benign
chr2	208325795	A	T	56.9	PASS	Benign	Benign
chr2	208325804	C	G	59.0	PASS	Benign	Benign
chr2	208326358	C	A	56.3	PASS	Benign	Benign
chr2	208326375	T	C	61.3	PASS	Benign	Benign
chr2	208347983	G	A	62.0	PASS	Benign	Benign
chr2	208350046	A	G	46.4	PASS	Benign	Benign
chr2	208350862	A	G	64.3	PASS	Benign	Benign

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr2	208320256	G	A	50.6	PASS	0.9932	0.991813
chr2	208320275	C	T	53.9	PASS	0.9312	0.911542
chr2	208325606	T	C	67.7	PASS	0.9309	0.911741
chr2	208325795	A	T	56.9	PASS	0.9305	0.911542
chr2	208325804	C	G	59.0	PASS	0.9311	0.912141
chr2	208326358	C	A	56.3	PASS	0.9310	0.911542
chr2	208326375	T	C	61.3	PASS	0.9956	0.996006
chr2	208347983	G	A	62.0	PASS	0.7192	0.716054
chr2	208350046	A	G	46.4	PASS	0.9305	0.911142
chr2	208350862	A	G	64.3	PASS	0.9306	0.911142

Name:	: John Doe	Patient ID	: ID20240320
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Fleck corneal dystrophy

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr2	208320256	G	A	non synonymous SNV	Autosomal dominant
chr2	208320275	C	T	synonymous SNV	Autosomal dominant
chr2	208325606	T	C	non synonymous SNV	Autosomal dominant
chr2	208325795	A	T	non synonymous SNV	Autosomal dominant
chr2	208325804	C	G	non synonymous SNV	Autosomal dominant
chr2	208326358	C	A	non synonymous SNV	Autosomal dominant
chr2	208326375	T	C	synonymous SNV	Autosomal dominant
chr2	208347983	G	A	synonymous SNV	Autosomal dominant
chr2	208350046	A	G	synonymous SNV	Autosomal dominant
chr2	208350862	A	G	synonymous SNV	Autosomal dominant

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Fleck corneal dystrophy

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr2	208320256	G	A	All ages	<1 / 1.000.000
chr2	208320275	C	T	All ages	<1 / 1.000.000
chr2	208325606	T	C	All ages	<1 / 1.000.000
chr2	208325795	A	T	All ages	<1 / 1.000.000
chr2	208325804	C	G	All ages	<1 / 1.000.000
chr2	208326358	C	A	All ages	<1 / 1.000.000
chr2	208326375	T	C	All ages	<1 / 1.000.000
chr2	208347983	G	A	All ages	<1 / 1.000.000
chr2	208350046	A	G	All ages	<1 / 1.000.000
chr2	208350862	A	G	All ages	<1 / 1.000.000

Name:	: John Doe	Patient ID	: ID20240320
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Hereditary breast cancer

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr2	214767531	C	T	24.7	QUAL30	Benign	Benign or Likely benign
chr2	214767532	A	G	24.0	QUAL30	Benign	Benign
chr2	214780740	C	G	32.0	PASS	Benign	Benign
chr5	163475510	T	C	22.2	QUAL30	Benign	Unknown
chr6	3010156	C	T	46.3	PASS	Benign	Unknown
chr6	3015556	A	G	60.0	PASS	Benign	Unknown
chr8	52624756	A	G	57.5	PASS	Benign	Unknown
chr8	52642509	A	G	59.8	PASS	Benign	Unknown
chr8	52657768	T	C	33.3	PASS	Likely benign	Benign
chr8	52674146	A	G	60.3	PASS	Benign	Unknown

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr2	214767531	C	T	24.7	QUAL30	0.3845	0.366214
chr2	214767532	A	G	24.0	QUAL30	0.7658	0.770567
chr2	214780740	C	G	32.0	PASS	0.5519	0.459265
chr5	163475510	T	C	22.2	QUAL30	0.2882	0.3127
chr6	3010156	C	T	46.3	PASS	0.8502	0.797524
chr6	3015556	A	G	60.0	PASS	0.7831	0.757388
chr8	52624756	A	G	57.5	PASS	0.1807	0.285343
chr8	52642509	A	G	59.8	PASS	0.1821	0.28754
chr8	52657768	T	C	33.3	PASS	0.0023	0.00858626
chr8	52674146	A	G	60.3	PASS	0.4460	0.502995

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Hereditary breast cancer

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr2	214767531	C	T	non synonymous SNV	-
chr2	214767532	A	G	synonymous SNV	-
chr2	214780740	C	G	non synonymous SNV	-
chr5	163475510	T	C	non synonymous SNV	-
chr6	3010156	C	T	non synonymous SNV	-
chr6	3015556	A	G	synonymous SNV	-
chr8	52624756	A	G	synonymous SNV	-
chr8	52642509	A	G	synonymous SNV	-
chr8	52657768	T	C	synonymous SNV	-
chr8	52674146	A	G	non synonymous SNV	-

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Hereditary breast cancer

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr2	214767531	C	T	-	-
chr2	214767532	A	G	-	-
chr2	214780740	C	G	-	-
chr5	163475510	T	C	-	-
chr6	3010156	C	T	-	-
chr6	3015556	A	G	-	-
chr8	52624756	A	G	-	-
chr8	52642509	A	G	-	-
chr8	52657768	T	C	-	-
chr8	52674146	A	G	-	-

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
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Adams-Oliver syndrome

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr3	68988937	A	G	29.2	QUAL30	Benign	Unknown
chr3	119409551	G	A	4.7	QUAL30	Benign	Unknown
chr3	119414336	G	A	68.6	PASS	Benign	Benign
chr3	119414707	G	A	25.6	QUAL30	Benign	Unknown
chr4	26415514	T	C	30.5	PASS	Benign	Unknown
chr9	136497184	G	A	67.1	PASS	Benign	Benign
chr9	136503255	G	A	59.3	PASS	Benign	Benign
chr9	136513480	A	G	53.4	PASS	Benign	Benign
chr9	136523808	A	G	53.2	PASS	Benign	Benign
chr19	11248123	G	A	31.8	PASS	Benign	Benign

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr3	68988937	A	G	29.2	QUAL30	0.1513	0.17492
chr3	119409551	G	A	4.7	QUAL30	0.1592	0.1877
chr3	119414336	G	A	68.6	PASS	0.8353	0.845647
chr3	119414707	G	A	25.6	QUAL30	0.1389	0.129792
chr4	26415514	T	C	30.5	PASS	0.5091	0.511781
chr9	136497184	G	A	67.1	PASS	0.5914	0.695088
chr9	136503255	G	A	59.3	PASS	0.4460	0.553514
chr9	136513480	A	G	53.4	PASS	0.4428	0.527756
chr9	136523808	A	G	53.2	PASS	0.6424	0.740016
chr19	11248123	G	A	31.8	PASS	0.5428	0.519569

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Adams-Oliver syndrome

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr3	68988937	A	G	synonymous SNV	Autosomal dominant, Autosomal recessive
chr3	119409551	G	A	synonymous SNV	Autosomal dominant, Autosomal recessive
chr3	119414336	G	A	non synonymous SNV	Autosomal dominant, Autosomal recessive
chr3	119414707	G	A	synonymous SNV	Autosomal dominant, Autosomal recessive
chr4	26415514	T	C	synonymous SNV	Autosomal dominant, Autosomal recessive
chr9	136497184	G	A	synonymous SNV	Autosomal dominant, Autosomal recessive
chr9	136503255	G	A	synonymous SNV	Autosomal dominant, Autosomal recessive
chr9	136513480	A	G	synonymous SNV	Autosomal dominant, Autosomal recessive
chr9	136523808	A	G	synonymous SNV	Autosomal dominant, Autosomal recessive
chr19	11248123	G	A	non synonymous SNV	Autosomal dominant, Autosomal recessive

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Adams-Oliver syndrome

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr3	68988937	A	G	Neonatal	Unknown
chr3	119409551	G	A	Neonatal	Unknown
chr3	119414336	G	A	Neonatal	Unknown
chr3	119414707	G	A	Neonatal	Unknown
chr4	26415514	T	C	Neonatal	Unknown
chr9	136497184	G	A	Neonatal	Unknown
chr9	136503255	G	A	Neonatal	Unknown
chr9	136513480	A	G	Neonatal	Unknown
chr9	136523808	A	G	Neonatal	Unknown
chr19	11248123	G	A	Neonatal	Unknown

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Amyotrophic lateral sclerosis

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr3	87227549	C	T	20.8	QUAL30	Benign	Benign
chr3	87245899	T	C	49.4	PASS	Benign	Benign
chr3	87249925	A	C	9.2	QUAL30	Benign	Benign
chr10	13110400	T	A	28.9	QUAL30	Benign	Benign or Likely benign
chr17	4945989	G	A	20.4	QUAL30	Benign	Benign
chr17	4946479	G	A	37.3	PASS	Benign	Unknown
chr22	29489484	C	T	22.3	QUAL30	Benign	Benign
chr22	29489578	A	AAAGT CCCCT GAGAA GGCC	29.8	QUAL30	Benign	Benign
chr22	29489872	T	C	7.9	QUAL30	Benign	Benign
chr22	29490424	A	G	22.9	QUAL30	Benign	Benign

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr3	87227549	C	T	20.8	QUAL30	0.0940	0.107228
chr3	87245899	T	C	49.4	PASS	0.8308	0.815695
chr3	87249925	A	C	9.2	QUAL30	0.0926	0.105831
chr10	13110400	T	A	28.9	QUAL30	0.0616	0.0786741
chr17	4945989	G	A	20.4	QUAL30	0.0444	0.0882588
chr17	4946479	G	A	37.3	PASS	0.5801	0.497404
chr22	29489484	C	T	22.3	QUAL30	0.1689	0.150559
chr22	29489578	A	AAAGT CCCCT GAGAA GGCC	29.8	QUAL30	0.6182	.
chr22	29489872	T	C	7.9	QUAL30	0.8237	0.818291
chr22	29490424	A	G	22.9	QUAL30	0.8264	0.850439

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Amyotrophic lateral sclerosis

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr3	87227549	C	T	synonymous SNV	Autosomal dominant, Autosomal recessive,
chr3	87245899	T	C	synonymous SNV	Autosomal dominant, Autosomal recessive,
chr3	87249925	A	C	synonymous SNV	Autosomal dominant, Autosomal recessive,
chr10	13110400	T	A	non synonymous SNV	Autosomal dominant, Autosomal recessive,
chr17	4945989	G	A	synonymous SNV	Autosomal dominant, Autosomal recessive,
chr17	4946479	G	A	synonymous SNV	Autosomal dominant, Autosomal recessive,
chr22	29489484	C	T	non synonymous SNV	Autosomal dominant, Autosomal recessive,
chr22	29489578	A	AAAGT CCCCT GAGAA GGCC	nonframeshift insertion	Autosomal dominant, Autosomal recessive,
chr22	29489872	T	C	synonymous SNV	Autosomal dominant, Autosomal recessive,
chr22	29490424	A	G	synonymous SNV	Autosomal dominant, Autosomal recessive,

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Amyotrophic lateral sclerosis

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr3	87227549	C	T	Adult	1-9 / 100 000
chr3	87245899	T	C	Adult	1-9 / 100 000
chr3	87249925	A	C	Adult	1-9 / 100 000
chr10	13110400	T	A	Adult	1-9 / 100 000
chr17	4945989	G	A	Adult	1-9 / 100 000
chr17	4946479	G	A	Adult	1-9 / 100 000
chr22	29489484	C	T	Adult	1-9 / 100 000
chr22	29489578	A	AAAGT CCCCT GAGAA GGCC	Adult	1-9 / 100 000
chr22	29489872	T	C	Adult	1-9 / 100 000
chr22	29490424	A	G	Adult	1-9 / 100 000

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Ellis Van Creveld syndrome

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr4	5568494	G	A	67.1	PASS	Benign	Benign
chr4	5622943	T	C	34.5	PASS	Benign	Benign
chr4	5689175	T	C	18.3	QUAL30	Benign	Benign
chr4	5708462	G	A	23.9	QUAL30	Benign	Benign
chr4	5741782	C	T	63.9	PASS	Benign	Benign
chr4	5741785	T	C	67.0	PASS	Benign	Benign
chr4	5748276	A	G	9.4	QUAL30	Benign	Benign
chr4	5753815	C	A	30.2	PASS	Benign	Benign
chr4	5783715	G	A	23.5	QUAL30	Benign	Benign
chr4	5793685	C	T	21.5	QUAL30	Benign	Benign

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr4	5568494	G	A	67.1	PASS	0.3037	0.440895
chr4	5622943	T	C	34.5	PASS	0.4506	0.517572
chr4	5689175	T	C	18.3	QUAL30	0.2180	0.265176
chr4	5708462	G	A	23.9	QUAL30	0.1711	0.21865
chr4	5741782	C	T	63.9	PASS	0.9533	0.966653
chr4	5741785	T	C	67.0	PASS	0.7357	0.744808
chr4	5748276	A	G	9.4	QUAL30	0.2638	0.217452
chr4	5753815	C	A	30.2	PASS	0.8332	0.844649
chr4	5783715	G	A	23.5	QUAL30	0.3227	0.301518
chr4	5793685	C	T	21.5	QUAL30	0.2662	0.333866

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Ellis Van Creveld syndrome

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr4	5568494	G	A	synonymous SNV	Autosomal recessive
chr4	5622943	T	C	non synonymous SNV	Autosomal recessive
chr4	5689175	T	C	non synonymous SNV	Autosomal recessive
chr4	5708462	G	A	non synonymous SNV	Autosomal recessive
chr4	5741782	C	T	synonymous SNV	Autosomal recessive
chr4	5741785	T	C	non synonymous SNV	Autosomal recessive
chr4	5748276	A	G	synonymous SNV	Autosomal recessive
chr4	5753815	C	A	non synonymous SNV	Autosomal recessive
chr4	5783715	G	A	non synonymous SNV	Autosomal recessive
chr4	5793685	C	T	synonymous SNV	Autosomal recessive

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Ellis Van Creveld syndrome

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr4	5568494	G	A	Neonatal, Antenatal	Unknown
chr4	5622943	T	C	Neonatal, Antenatal	Unknown
chr4	5689175	T	C	Neonatal, Antenatal	Unknown
chr4	5708462	G	A	Neonatal, Antenatal	Unknown
chr4	5741782	C	T	Neonatal, Antenatal	Unknown
chr4	5741785	T	C	Neonatal, Antenatal	Unknown
chr4	5748276	A	G	Neonatal, Antenatal	Unknown
chr4	5753815	C	A	Neonatal, Antenatal	Unknown
chr4	5783715	G	A	Neonatal, Antenatal	Unknown
chr4	5793685	C	T	Neonatal, Antenatal	Unknown

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Osteogenesis imperfecta type 3

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr7	94413927	C	G	65.6	PASS	Benign	Benign
chr11	75566583	A	G	31.2	PASS	Benign	Benign
chr11	75566712	C	G	28.5	QUAL30	Benign	Benign
chr11	75568801	C	T	25.8	QUAL30	Benign	Benign
chr11	75571837	G	A	35.4	PASS	Benign	Benign
chr17	1769982	C	T	38.7	PASS	Benign	Benign
chr17	1771135	T	C	14.9	QUAL30	Benign	Benign
chr17	1776708	T	C	60.2	PASS	Benign	Benign
chr17	50190862	A	G	60.7	PASS	Benign	Benign
chr17	50199235	G	A	23.7	QUAL30	Likely benign	Conflicting

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr7	94413927	C	G	65.6	PASS	0.8092	0.821685
chr11	75566583	A	G	31.2	PASS	0.9125	0.889976
chr11	75566712	C	G	28.5	QUAL30	0.3610	0.373003
chr11	75568801	C	T	25.8	QUAL30	0.3638	0.376997
chr11	75571837	G	A	35.4	PASS	0.3627	0.376797
chr17	1769982	C	T	38.7	PASS	0.6744	0.626198
chr17	1771135	T	C	14.9	QUAL30	0.2837	0.300319
chr17	1776708	T	C	60.2	PASS	0.7697	0.776757
chr17	50190862	A	G	60.7	PASS	0.9996	0.9998
chr17	50199235	G	A	23.7	QUAL30	0.0004	0.00159744

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Osteogenesis imperfecta type 3

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr7	94413927	C	G	non synonymous SNV	Autosomal dominant, Autosomal recessive
chr11	75566583	A	G	synonymous SNV	Autosomal dominant, Autosomal recessive
chr11	75566712	C	G	synonymous SNV	Autosomal dominant, Autosomal recessive
chr11	75568801	C	T	synonymous SNV	Autosomal dominant, Autosomal recessive
chr11	75571837	G	A	synonymous SNV	Autosomal dominant, Autosomal recessive
chr17	1769982	C	T	non synonymous SNV	Autosomal dominant, Autosomal recessive
chr17	1771135	T	C	synonymous SNV	Autosomal dominant, Autosomal recessive
chr17	1776708	T	C	synonymous SNV	Autosomal dominant, Autosomal recessive
chr17	50190862	A	G	synonymous SNV	Autosomal dominant, Autosomal recessive
chr17	50199235	G	A	synonymous SNV	Autosomal dominant, Autosomal recessive

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Osteogenesis imperfecta type 3

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr7	94413927	C	G	Infancy, Neonatal	Unknown
chr11	75566583	A	G	Infancy, Neonatal	Unknown
chr11	75566712	C	G	Infancy, Neonatal	Unknown
chr11	75568801	C	T	Infancy, Neonatal	Unknown
chr11	75571837	G	A	Infancy, Neonatal	Unknown
chr17	1769982	C	T	Infancy, Neonatal	Unknown
chr17	1771135	T	C	Infancy, Neonatal	Unknown
chr17	1776708	T	C	Infancy, Neonatal	Unknown
chr17	50190862	A	G	Infancy, Neonatal	Unknown
chr17	50199235	G	A	Infancy, Neonatal	Unknown

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Localized junctional epidermolysis bullosa, non-Herlitz type

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr10	104033992	T	C	29.9	QUAL30	Benign	Benign
chr10	104037061	A	G	57.5	PASS	Benign	Benign
chr10	104039114	T	C	57.4	PASS	Benign	Benign
chr10	104039458	G	T	61.8	PASS	Benign	Benign
chr10	104040408	T	A	15.9	QUAL30	Likely benign	Benign
chr10	104041495	G	A	67.6	PASS	Benign	Benign
chr10	104050642	T	C	23.2	QUAL30	Benign	Benign
chr10	104057158	C	T	50.0	PASS	Benign	Benign
chr10	104060198	G	A	14.0	QUAL30	Benign	Benign
chr10	104064575	G	A	28.7	QUAL30	Benign	Benign

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr10	104033992	T	C	29.9	QUAL30	0.2789	0.258586
chr10	104037061	A	G	57.5	PASS	0.9874	0.986022
chr10	104039114	T	C	57.4	PASS	0.7260	0.68151
chr10	104039458	G	T	61.8	PASS	0.7228	0.683506
chr10	104040408	T	A	15.9	QUAL30	0.0008	0.00279553
chr10	104041495	G	A	67.6	PASS	0.7221	0.673922
chr10	104050642	T	C	23.2	QUAL30	0.7248	0.69369
chr10	104057158	C	T	50.0	PASS	0.8533	0.81849
chr10	104060198	G	A	14.0	QUAL30	0.5766	0.552516
chr10	104064575	G	A	28.7	QUAL30	0.6014	0.564896

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Localized junctional epidermolysis bullosa, non-Herlitz type

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr10	104033992	T	C	non synonymous SNV	Autosomal recessive
chr10	104037061	A	G	synonymous SNV	Autosomal recessive
chr10	104039114	T	C	synonymous SNV	Autosomal recessive
chr10	104039458	G	T	synonymous SNV	Autosomal recessive
chr10	104040408	T	A	non synonymous SNV	Autosomal recessive
chr10	104041495	G	A	synonymous SNV	Autosomal recessive
chr10	104050642	T	C	non synonymous SNV	Autosomal recessive
chr10	104057158	C	T	non synonymous SNV	Autosomal recessive
chr10	104060198	G	A	synonymous SNV	Autosomal recessive
chr10	104064575	G	A	non synonymous SNV	Autosomal recessive

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Localized junctional epidermolysis bullosa, non-Herlitz type

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr10	104033992	T	C	Infancy, Neonatal	<1 / 1.000.000
chr10	104037061	A	G	Infancy, Neonatal	<1 / 1.000.000
chr10	104039114	T	C	Infancy, Neonatal	<1 / 1.000.000
chr10	104039458	G	T	Infancy, Neonatal	<1 / 1.000.000
chr10	104040408	T	A	Infancy, Neonatal	<1 / 1.000.000
chr10	104041495	G	A	Infancy, Neonatal	<1 / 1.000.000
chr10	104050642	T	C	Infancy, Neonatal	<1 / 1.000.000
chr10	104057158	C	T	Infancy, Neonatal	<1 / 1.000.000
chr10	104060198	G	A	Infancy, Neonatal	<1 / 1.000.000
chr10	104064575	G	A	Infancy, Neonatal	<1 / 1.000.000

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Porencephaly

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr13	110424850	G	A	62.3	PASS	Benign	Benign
chr13	110445879	C	T	67.5	PASS	Benign	Benign
chr13	110449695	G	A	15.3	QUAL30	Benign	Benign
chr13	110449779	C	T	18.3	QUAL30	Benign	Benign
chr13	110501711	T	A	60.3	PASS	Benign	Unknown
chr13	110501714	T	C	56.4	PASS	Benign	Unknown
chr13	110503426	T	C	58.3	PASS	Benign	Unknown
chr13	110503432	G	A	29.7	QUAL30	Benign	Benign
chr13	110504152	C	T	63.8	PASS	Benign	Unknown
chr13	110506527	A	G	22.2	QUAL30	Benign	Benign

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr13	110424850	G	A	62.3	PASS	0.9382	0.942692
chr13	110445879	C	T	67.5	PASS	0.4947	0.482827
chr13	110449695	G	A	15.3	QUAL30	0.3057	0.238618
chr13	110449779	C	T	18.3	QUAL30	0.3275	0.246805
chr13	110501711	T	A	60.3	PASS	0.9470	0.939497
chr13	110501714	T	C	56.4	PASS	0.9469	0.939497
chr13	110503426	T	C	58.3	PASS	0.9852	0.980232
chr13	110503432	G	A	29.7	QUAL30	0.2190	0.186302
chr13	110504152	C	T	63.8	PASS	0.9454	0.935903
chr13	110506527	A	G	22.2	QUAL30	0.8339	0.751997

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Porencephaly

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr13	110424850	G	A	synonymous SNV	Multigenic/multifactorial,
chr13	110445879	C	T	synonymous SNV	Multigenic/multifactorial,
chr13	110449695	G	A	synonymous SNV	Multigenic/multifactorial,
chr13	110449779	C	T	synonymous SNV	Multigenic/multifactorial,
chr13	110501711	T	A	synonymous SNV	Multigenic/multifactorial,
chr13	110501714	T	C	synonymous SNV	Multigenic/multifactorial,
chr13	110503426	T	C	synonymous SNV	Multigenic/multifactorial,
chr13	110503432	G	A	synonymous SNV	Multigenic/multifactorial,
chr13	110504152	C	T	synonymous SNV	Multigenic/multifactorial,
chr13	110506527	A	G	synonymous SNV	Multigenic/multifactorial,

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Porencephaly

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr13	110424850	G	A	Infancy, Neonatal	Unknown
chr13	110445879	C	T	Infancy, Neonatal	Unknown
chr13	110449695	G	A	Infancy, Neonatal	Unknown
chr13	110449779	C	T	Infancy, Neonatal	Unknown
chr13	110501711	T	A	Infancy, Neonatal	Unknown
chr13	110501714	T	C	Infancy, Neonatal	Unknown
chr13	110503426	T	C	Infancy, Neonatal	Unknown
chr13	110503432	G	A	Infancy, Neonatal	Unknown
chr13	110504152	C	T	Infancy, Neonatal	Unknown
chr13	110506527	A	G	Infancy, Neonatal	Unknown

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Spondyloepimetaphyseal dysplasia, aggrecan type

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr15	88847929	C	A	22.6	QUAL30	Benign	Unknown
chr15	88849514	T	C	59.3	PASS	Benign	Benign
chr15	88855099	G	A	32.2	PASS	Benign	Unknown
chr15	88855322	C	A	30.2	PASS	Benign	Unknown
chr15	88857878	A	G	20.8	QUAL30	Benign	Unknown
chr15	88858148	C	T	22.8	QUAL30	Likely benign	Unknown
chr15	88858384	T	G	59.6	PASS	Benign	Unknown
chr15	88858820	A	G	60.0	PASS	Benign	Unknown
chr15	88859008	T	C	66.4	PASS	Benign	Unknown
chr15	88859365	T	G	10.8	QUAL30	Benign	Unknown

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr15	88847929	C	A	22.6	QUAL30	0.7521	0.694289
chr15	88849514	T	C	59.3	PASS	0.9885	0.995607
chr15	88855099	G	A	32.2	PASS	0.8139	0.758986
chr15	88855322	C	A	30.2	PASS	0.0811	0.0972444
chr15	88857878	A	G	20.8	QUAL30	0.8151	0.760383
chr15	88858148	C	T	22.8	QUAL30	0.0051	0.0147764
chr15	88858384	T	G	59.6	PASS	0.6195	0.532548
chr15	88858820	A	G	60.0	PASS	0.7074	0.645966
chr15	88859008	T	C	66.4	PASS	0.7805	0.736422
chr15	88859365	T	G	10.8	QUAL30	0.5293	0.451478

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Spondyloepimetaphyseal dysplasia, aggrecan type

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr15	88847929	C	A	synonymous SNV	Autosomal recessive
chr15	88849514	T	C	synonymous SNV	Autosomal recessive
chr15	88855099	G	A	synonymous SNV	Autosomal recessive
chr15	88855322	C	A	non synonymous SNV	Autosomal recessive
chr15	88857878	A	G	non synonymous SNV	Autosomal recessive
chr15	88858148	C	T	non synonymous SNV	Autosomal recessive
chr15	88858384	T	G	synonymous SNV	Autosomal recessive
chr15	88858820	A	G	non synonymous SNV	Autosomal recessive
chr15	88859008	T	C	synonymous SNV	Autosomal recessive
chr15	88859365	T	G	synonymous SNV	Autosomal recessive

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Spondyloepimetaphyseal dysplasia, aggrecan type

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr15	88847929	C	A	Infancy, Neonatal	<1 / 1.000.000
chr15	88849514	T	C	Infancy, Neonatal	<1 / 1.000.000
chr15	88855099	G	A	Infancy, Neonatal	<1 / 1.000.000
chr15	88855322	C	A	Infancy, Neonatal	<1 / 1.000.000
chr15	88857878	A	G	Infancy, Neonatal	<1 / 1.000.000
chr15	88858148	C	T	Infancy, Neonatal	<1 / 1.000.000
chr15	88858384	T	G	Infancy, Neonatal	<1 / 1.000.000
chr15	88858820	A	G	Infancy, Neonatal	<1 / 1.000.000
chr15	88859008	T	C	Infancy, Neonatal	<1 / 1.000.000
chr15	88859365	T	G	Infancy, Neonatal	<1 / 1.000.000

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Glycogen storage disease due to acid maltase deficiency

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr17	80104910	T	C	48.9	PASS	Benign	Benign
chr17	80105798	A	G	56.0	PASS	Benign	Benign
chr17	80105870	G	A	60.7	PASS	Benign	Benign
chr17	80108705	G	A	51.7	PASS	Benign	Benign
chr17	80110970	G	A	34.2	PASS	Benign	Benign
chr17	80112072	G	A	27.2	QUAL30	Likely benign	Benign, other
chr17	80113242	G	A	21.5	QUAL30	Benign	Benign, other
chr17	80113310	A	G	25.1	QUAL30	Benign	Benign
chr17	80117606	G	A	65.9	PASS	Benign	Benign
chr17	80118264	G	A	14.3	QUAL30	Benign	Benign

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr17	80104910	T	C	48.9	PASS	0.7276	0.714457
chr17	80105798	A	G	56.0	PASS	0.6677	0.600839
chr17	80105870	G	A	60.7	PASS	0.6676	0.602436
chr17	80108705	G	A	51.7	PASS	0.6654	0.602835
chr17	80110970	G	A	34.2	PASS	0.1972	0.159545
chr17	80112072	G	A	27.2	QUAL30	0.0121	0.0371406
chr17	80113242	G	A	21.5	QUAL30	0.0532	0.0780751
chr17	80113310	A	G	25.1	QUAL30	0.2537	0.241613
chr17	80117606	G	A	65.9	PASS	0.7265	0.711861
chr17	80118264	G	A	14.3	QUAL30	0.5779	0.509385

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Glycogen storage disease due to acid maltase deficiency

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr17	80104910	T	C	synonymous SNV	Autosomal recessive
chr17	80105798	A	G	non synonymous SNV	Autosomal recessive
chr17	80105870	G	A	non synonymous SNV	Autosomal recessive
chr17	80108705	G	A	synonymous SNV	Autosomal recessive
chr17	80110970	G	A	synonymous SNV	Autosomal recessive
chr17	80112072	G	A	non synonymous SNV	Autosomal recessive
chr17	80113242	G	A	non synonymous SNV	Autosomal recessive
chr17	80113310	A	G	synonymous SNV	Autosomal recessive
chr17	80117606	G	A	non synonymous SNV	Autosomal recessive
chr17	80118264	G	A	synonymous SNV	Autosomal recessive

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Glycogen storage disease due to acid maltase deficiency

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr17	80104910	T	C	Antenatal, Neonatal, Infancy, Childhood, Adolescent, Adult	Unknown
chr17	80105798	A	G	Antenatal, Neonatal, Infancy, Childhood, Adolescent, Adult	Unknown
chr17	80105870	G	A	Antenatal, Neonatal, Infancy, Childhood, Adolescent, Adult	Unknown
chr17	80108705	G	A	Antenatal, Neonatal, Infancy, Childhood, Adolescent, Adult	Unknown
chr17	80110970	G	A	Antenatal, Neonatal, Infancy, Childhood, Adolescent, Adult	Unknown
chr17	80112072	G	A	Antenatal, Neonatal, Infancy, Childhood, Adolescent, Adult	Unknown
chr17	80113242	G	A	Antenatal, Neonatal, Infancy, Childhood, Adolescent, Adult	Unknown

Name: : John Doe
Gender : Male
Date of Birth : 01/01/1990

Patient ID : ID20240320
Ethnicity : Javanese

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr17	80113310	A	G	Antenatal, Neonatal, Infancy, Childhood, Adolescent, Adult	Unknown
chr17	80117606	G	A	Antenatal, Neonatal, Infancy, Childhood, Adolescent, Adult	Unknown
chr17	80118264	G	A	Antenatal, Neonatal, Infancy, Childhood, Adolescent, Adult	Unknown

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome

Variant Classification and Significance

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	ACMG Classification	ClinVar Significance
chr22	25763322	G	A	25.8	QUAL30	Benign	Unknown
chr22	25770933	G	C	65.9	PASS	Benign	Unknown
chr22	25777694	T	C	59.3	PASS	Benign	Unknown
chr22	25798015	C	G	16.5	QUAL30	Benign	Unknown
chr22	25798078	T	C	27.6	QUAL30	Benign	Unknown
chr22	25823638	G	A	13.5	QUAL30	Benign	Unknown
chr22	25843883	C	A	56.1	PASS	Benign	Unknown
chr22	25851489	G	A	34.7	PASS	Benign	Unknown
chr22	26026488	T	C	41.5	PASS	Likely benign	Unknown
chr22	26027014	A	G	29.9	QUAL30	Benign	Unknown

Variant Population Frequency

Chromosome	Position	Reference Allele	Alternate Allele	Variant Quality	Filter Status	gnomAD AF	1000g AF
chr22	25763322	G	A	25.8	QUAL30	0.5802	0.701677
chr22	25770933	G	C	65.9	PASS	0.9730	0.938498
chr22	25777694	T	C	59.3	PASS	0.9680	0.932508
chr22	25798015	C	G	16.5	QUAL30	0.6554	0.684305
chr22	25798078	T	C	27.6	QUAL30	0.5952	0.56889
chr22	25823638	G	A	13.5	QUAL30	0.2639	0.159145
chr22	25843883	C	A	56.1	PASS	0.6653	0.723442
chr22	25851489	G	A	34.7	PASS	0.3211	0.242013
chr22	26026488	T	C	41.5	PASS	0.0039	0.0301518
chr22	26027014	A	G	29.9	QUAL30	0.3534	0.426318

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome

Variant Consequence and Inheritance Pattern

Chromosome	Position	Reference Allele	Alternate Allele	Consequence	Inheritance Pattern
chr22	25763322	G	A	non synonymous SNV	Autosomal recessive
chr22	25770933	G	C	non synonymous SNV	Autosomal recessive
chr22	25777694	T	C	non synonymous SNV	Autosomal recessive
chr22	25798015	C	G	synonymous SNV	Autosomal recessive
chr22	25798078	T	C	synonymous SNV	Autosomal recessive
chr22	25823638	G	A	synonymous SNV	Autosomal recessive
chr22	25843883	C	A	non synonymous SNV	Autosomal recessive
chr22	25851489	G	A	synonymous SNV	Autosomal recessive
chr22	26026488	T	C	synonymous SNV	Autosomal recessive
chr22	26027014	A	G	non synonymous SNV	Autosomal recessive

Name:	: John Doe	Patient ID	: ID20240320
Gender	: Male	Ethnicity	: Javanese
Date of Birth	: 01/01/1990		

Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome

Variant Age of Onset and Prevalence

Chromosome	Position	Reference Allele	Alternate Allele	Age of Onset	Prevalence
chr22	25763322	G	A	Infancy	<1 / 1.000.000
chr22	25770933	G	C	Infancy	<1 / 1.000.000
chr22	25777694	T	C	Infancy	<1 / 1.000.000
chr22	25798015	C	G	Infancy	<1 / 1.000.000
chr22	25798078	T	C	Infancy	<1 / 1.000.000
chr22	25823638	G	A	Infancy	<1 / 1.000.000
chr22	25843883	C	A	Infancy	<1 / 1.000.000
chr22	25851489	G	A	Infancy	<1 / 1.000.000
chr22	26026488	T	C	Infancy	<1 / 1.000.000
chr22	26027014	A	G	Infancy	<1 / 1.000.000