

**Genetics Research. The prevalence of clinical features in patients with Aarsk**

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**Supplementary Table S1: L**

Authors	Year	Country	Patient	Age(years)	Phenotypes
Schwartz et al. [3]	2000	USA§	1(FC)	7	Brachydactyly/wide fingers; Cupid's Bow; Dorsal Groove penis; Downward Slant Palpebral Fissures; Hypertelorism ; Interphalangeal Joint Contracture; Joint Hypermobility; Long Philtrum; Macrocephaly; Pointed Chin.; Ptosis; Short Stature; Simian Creases; Umbilical Hernia
			2(IT)	12	Brachydactyly/wide fingers; Cupid's Bow; Downward Slanted Palpebral Fissures; Hypertelorism; Inguinal Hernia; Interphalangeal Joint Contracture; Joint hypermobility; Long Philtrum; Macrocephaly; Pointed Chin; Ptosis; Shawl Scrotum; Short Stature; Simian Crease
			3(MK)	11	Anteverted Nostrils; Brachydactyly/Wide Fingers; Hypertelorism; Shawl Scrotum; Short Stature; Syndactyly; Cryptorchidism; Downward slant palpebral fissures; Inguinal hernia; Ptosis; Widow's peak; Bone Age retarded; Myopia

Orrico et al. [37]	2000	Italy	4(II1)	66	Anteverted Nostrils; Camptodactyly; Clinodactyly; Crease Below Lower Lip; Cryptorchidism; Downward Slanted Palpebral Fissures; Hypertelorism; Inguinal Hernia; Interdigital Webbing; Ptosis; Scrotum; Short Stature; Short/Broad Hands; Umbilical Hernia; Widow's Peak
			5(IV9)	9	Anteverted Nostrils; Anteverted nostrils/Short Nose; Clinodactyly; Crease Below Lower Lip; Cryptorchidism; Downward Slant Palpebral Fissures; Dysplastic Ears; Frontal Bossing; Hypertelorism; Inguinal Hernia; Interdigital Webbing; Joint Hypermobility; Long Philtrum; Midface Hypoplasia; Ptosis; Round Face; Scrotum; Short Stature; Short/Broad Hands; Umbilical Hernia; Wide feet; Widow's peak
			6(IV10)	19	Anteverted Nostrils; Anteverted nostrils/Short Nose; Clinodactyly; Crease Below Lower Lip; Cryptorchidism; Downward Slant Palpebral Fissures; Hypertelorism; Inguinal Hernia; Interdigital Webbing; Joint Hypermobility; Ptosis; Scrotum; Short Stature; Short/Broad Hands; Umbilical Hernia; Widow's Peak

		NR*	7(50)	29	Anteverted Nostrils; Anteverted nostrils/Short Nose; Brachydactyly/Wide Fingers; Camptodactyly; Clinodactyly; Cryptorchidism; Hydrocele; Hypertelorism; Inguinal Hernia; Joint Hypermobility; Long Philtrum; Oligoasthenozoospermia; Ptosis; Shawl Scrotum; Short Stature; Syndactyly
		NR*	8(90)	9	Anteverted Nostrils; Brachydactyly/Wide Fingers; Camptodactyly; Cryptorchidism; Downward Slant Palpebral Fissures; Dysplastic Ears; Frontal Bossing; Hypertelorism; Inguinal Hernia; Interdigital Webbing; Joint Hypermobility; Long Philtrum; Ptosis; Round Face; Shawl Scrotum; Short Stature; Short/Broad Hands; Syndactyly; Widow's Peak
		Portugal	9(25)	11	Anteverted Nostrils; Brachydactyly/Wide Fingers; Camptodactyly; Cryptorchidism; Downward Slant Palpebral Fissures; Dysplastic Ears; Frontal Bossing; Hypertelorism; Inguinal Hernia; Interdigital Webbing; Joint Hypermobility; Long Philtrum; Ptosis; Round Face; Shawl Scrotum; Short Stature; Short/Broad Hands; Syndactyly; Widow's Peak

Orrico et al. [6]	2004	NR*	10(53)	1.3	Anteverted nostrils/Short Nose; Brachydactyly/wide fingers; Clinodactyly; Cryptorchidism; Downward slant palpebral fissures; Hypertelorism; Inguinal hernia ; Joint hypermobility; Long Philtrum;Shawl Scrotum ;Short stature; Simian Crease; Syndactyly; Umbilical hernia; Widow's peak
		NR*	11(61)	21	Behavioral Disorders;Brachydactyly/wide fingers; Camptodactyly; Hypertelorism; Joint hypermobility;Ptosis; Shawl Scrotum; short neck; Short stature; Social/Language skills delayed; Strabismus; Syndactyly
		NR*	12(26)	3	Anteverted nostrils/Short Nose;Anteverted nostrils/Short Nose; Brachydactyly/wide fingers; Clinodactyly; Epicanthal fold; Hypertelorism ; Inguinal hernia; Joint hypermobility; Long philtrum; Metatarsus varus; Round face; Shawl Scrotum; Short stature;Syndactyly; Umbilical hernia; Widow's peak
		NR*	13(73)	4.5	Anteverted nostrils/Short Nose; Bone age retarded; Camptodactyly; Cleft Lip Palate; Downward slant palpebral fissures; Epicanthal fold; Hypertelorism; Inguinal Hernia; Joint Hypermobility; Long Philtrum; Low set ears; Ptosis; Shawl Scrotum; Syndactyly; Widow's peak

		NR*	14(58)	1.6	ADHD; Anteverted nostrils/Short Nose; Axial Hypotonia; Brachydactyly/Wide fingers; Cryptorchidism; Development delayed; Hypertelorism; Joint Hypermobility; Long Philtrum; Low set ears; Prominent umbilicus; Ptosis; Shawl Scrotum; Short Stature; Syndactyly
		Ireland	15(65)	16	Anteverted nostrils/Short Nose; Brachydactyly/Wide Fingers; Camptodactyly; Cryptorchidism; Downward slant palpebral fissures; Hypertelorism; Long philtrum; Ptosis; Shawl Scrotum; Short/Broad Hands; Syndactyly; Wide feet
<b>Orrico et al. [7]</b>	2005	Italy	16	16	ADHD; Axial hypotonia; Behavioral disorder; Borderline mental retardation; Brachydactyly/Wide fingers; Broad NA†sal bridge; Downward slant palpebral fissures; Dysplastic ears; Enlarged Virchow-Robin spaces; HyperNA†sal speech; Hypertelorism; Interdigital webbing; Joint hypermobility; Low set ears; MicrogNA†thia; Midface Hypoplasia; Ogival palate; Pectus excavatum; Prominent umbilicus; ptosis; Short stature; Simian Creases; Strabismus

Shalev et al. [24]	2006	Israel	17(IV2)	24	Anteverted nostrils/Short Nose; Brachydactyly/Wide fingers; Camptodactyly; Dysplastic ears; Hypertelorism; Long philtrum; Pectus carinatum; Ptosis; Round face; Shawl Scrotum Short stature; Simian creases; Syndactyly
			18(IV3)	16	Brachydactyly/wide finger; Camptodactyly; Dysplastic ears; Frontal bossing; Hypertelorism; Ptosis; Round face; Short Stature; Simian creases; Syndactyly
			19(IV4)	2	Anteverted nostrils/Short Nose; Behavioral disorder; Development delayed; Fine and gross motor skills delayed; Hypertelorism; Microcephaly, Neurocognitive development delayed; Ptosis; Seizures; Short stature; Social/Language Skills delayed; Vertigo
KaNAjme et al. [8]	2006	Japan	20(1)	13	ADHD;Brachydactyly/Wide fingers; Frontal bossing; Hypertelorism; Interdigital webbing; Interphalangeal joint contracture; Joint hypermobility; Ptosis; Shawl scrotum; Short stature; Short/Broad Hands; Wide feet
			21(2)	4	Brachydactyly/Wide fingers; Frontal Bossing; Hypertelorism; Interdigital Webbing; Interphalangeal joint contracture; Joint hypermobility; Shawl Scrotum; Short stature; Short/Broad Hands; wide feet

<b>Bottani et al. [25]</b>	2007	Switzerland	22	8	Clinodactyly; Clubbed feet; Cryptorchidism; Inguinal hernia; Joint hypermobility; Pectus excavatum; Seizures; Shawl Scrotum; Unilateral focal frontoparietal PMG, Migraine headaches
<b>Orrico et al. [26]</b>	2007	Italy	23	15	Agenesis of Corpus Callosum; Anteverted nostrils/Short Nose; Brachydactyly/wide fingers; Crease below lower lip; Downward slant palpebral fissures; Dysplastic ears; Fine and gross motor skills delayed; Hydrocele; Hypertelorism; Inguinal hernia; Interdigital webbing; Long philtrum; Midface Hypoplasia; Neurocognitive development delayed; Omphalocele; Parietal-occipital encephalocele; Pectus excavatum; Ptosis; Right hemifacial microsomia; Shawl Scrotum; Short neck; Short stature; Short/Broad Hands; Simian Creases; still's murmur; Submucous cleft palate; Synophry; Transethmoidal; Meningoencephalocele; Ventricular septal defect; Wide feet; Widow's Peak
			24	6	Anteverted nostrils/Short Nose; Bifid uvula; Brachydactyly/Wide fingers; Camptodactyly; Crease below lower lip; Inguinal hernia; Interdigital webbing; Long philtrum; Low set ears; Metatarsus varus; Pectus excavatum; Prominent umbilicus; Ptosis; Round face; Shawl Scrotum; Short neck; Short stature; Simian Creases; Synophrys; Telecanthus; Widow's peak

<p><b>Bedoyan et al. [27]</b></p>	<p>2009</p>	<p>USAŞ</p>	<p>25</p>	<p>1.25</p>	<p>Anteverted nostrils/Short Nose; Astigmatism; Camptodactyly; Cryptorchidism; Downward slant palpebral fissures; Dysplastic ears; Hypertelorism; Inguinal hernia; Ogival palate; Ptosis; Shawl Scrotum</p>
		<p>NR*</p>	<p>26(9)</p>	<p>NR*</p>	<p>Anteverted nostrils; Brachydactyly/Wide fingers; Clinodactyly; Cryptorchidism; Dental malocclusion; Dysplastic ears; Hypertelorism; long philtrum; Midface hypoplasia; Obesity; Shawl scrotum; Short stature; Short/Broad Hands; Wide feet</p>
		<p>NR*</p>	<p>27(2)</p>	<p>NR*</p>	<p>Anteverted nostrils/Short Nose; Bone age retarded; Development delayed; Dysplastic ears; Hypertelorism; Obesity; preauricular tag; Shawl scrotum; Short stature; Short/Short/Broad Hands; Simian creases; Widow's peak</p>
		<p>NR*</p>	<p>28(5)</p>	<p>NR*</p>	<p>Anteverted nostrils/Short Nose; Camptodactyly; Cryptorchidism; Development delayed; Hypertelorism; Inguinal hernia; Interdigital webbing; Long philtrum; Metatarsus varus; Ptosis; Shawl scrotum; Short stature; Short/Broad Hands; Simian creases; Umbilical hernia; Wide feet; Widow's peak</p>



Orrico et al. [16]

2010

NR*	29(10)	NR*	Anteverted nostrils/Short Nose; Brachydactyly/wide fingers; Camptodactyly; Clinodactyly; Crease below lower lip; Downward slant palpebral fissures; Fetal Nuchal Oedema; Frontal bossing; Hypertelorism; Interdigital webbing; Long philtrum; Metatarsus varus; Microcephaly; Ptosis; Short stature; Short/Broad Hands; Simian creases; Wide feet
NR*	30(3)	NR*	Anteverted nostrils/Short Nose; Brachycephaly; Brachydactyly/wide fingers; Clinodactyly; Crease below lower lip; Development delayed; Dysplastic ears; Hypertelorism; Hypospadias; Inguinal hernia; Long Philtrum; Micropenis; Midface hypoplasia; Obesity; Ptosis; Shawl scrotum, Short stature; Short/Short/Broad Hands; Umbilical hernia; Widow's peak
NR*	31(11)	NR*	Anteverted nostrils/Short Nose; bone age retarded; Brachydactyly/Wide fingers; Clinodactyly; crease below lower lip; downward slant palpebral fissures; frontal bossing; Hypertelorism; long philtrum; midface hypoplasia; Patent ductus arteriosus; prominent umbilicus; ptosis; shawl scrotum; Short Stature; Short/Broad Hands; wide feet; widow's peak
NR*	32(8)	NR*	Anteverted nostrils/Short Nose; Anteverted nostrils/Short Nose; Brachydactyly/wide fingers; Camptodactyly; Clinodactyly; Cryptorchidism; Hypertelorism; long philtrum; obesity; Shawl scrotum; Short stature; Short/Broad Hands; Wide feet

		NR*	33(6)	NR*	Anteverted nostrils/Short Nose; bone age retarded; Brachydactyly/Wide fingers; Camptodactyly; clinodactyly; crease below lower lip; cryptorchidism; downward slant palpebral fissures; dysplastic ears; hypertelorism; joint hypermobility; shawl scrotum; Short stature; Short/Broad Hands; simian creases; Strabismus; wide feet; widow's peak
		NR*	34(7)	NR*	Anteverted nostrils/Short Nose; Brachydactyly/wide fingers; Clinodactyly; Congenital glaucoma; crease below lower lip; cryptorchidism; Dysplastic ears; frontal bossing; hypertelorism; Interdigital webbing; Joint hypermobility; long philtrum; metatarsus varus; midface hypoplasia; Shawl scrotum; short stature; Short/Broad Hands; Wide feet; Widow's peak
		NR*	35(1)	NR*	Anteverted nostrils/Short Nose; Anteverted nostrils/Short Nose; bone age retarded; crease below lower lip; downward slant palpebral fissures; dysplastic ears; Exostosis; hypertelorism; large eyebrows; long philtrum; pectus excavatum; shawl scrotum; short stature; Short/Broad Hands; wide feet; widow's peak
		NR*	36(4)	NR*	Anteverted nostrils/Short Nose; Brachydactyly/wide fingers; camptodactyly; clinodactyly; clubbed feet; Cryptorchidism; development delayed; Frontal bossing; hypertelorism; interdigital webbing; Shawl Scrotum; Short stature; Short/Broad Hands; Widow's peak

Pillozzi-Edmonds et al. [28]	2011	Canada	37(1)	0.75	Axial hypotonia; bitemporal narrowing; Clinodactyly; clubbed feet; frontal bossing; hydronephrosis; hypertelorism; inguinal hernia; interphalangeal joint contractures; low set ears; preauricular tag; Shawl scrotum; simian creases; vesicoureteral reflux
			38(2)	0.75	Anteverted nostrils/Short Nose; Axial Hypotonia; broad nasal bridge; clinodactyly; clubbed feet; cryptorchidism; frontal bossing; hypertelorism; interphalangeal joint contracture; long philtrum; low set ears; myopia; preauricular tag; Shawl Scrotum. Inguinal Hernia; Simian Creases; thin upper lip vermilion; triangular downturned mouth; upslanted palpebral fissures; vesicoureteral reflux
Ronce et al. [29]	2012	France	39	NR*	Anteverted nostrils/Short Nose; arthrogryposis; Camptodactyly; cryptorchidism; dysplastic ears; hypertelorism; joint hypermobility; large fontanelle/sutures; long philtrum; low set ears; metatarsus varus; micropenis; midface hypoplasia; Retrognathia; round face; short neck; Short/Broad Hands; simian creases; wide feet; widow's peak
			40(III-1)	NR*	Anteverted nostrils/Short Nose; cleft lip palate; cow's lick; cryptorchidism; dysplastic ears; Hypertelorism; interphalangeal joint contracture; joint hypermobility; long philtrum; shawl scrotum; short stature; Short/Broad Hands; wide feet

<b>Aten et al. [30]</b>	2012	Netherlands	41 (III-2)	NR*	Anteverted nostrils/Short Nose; cow's lick; cryptorchidism; dysplastic ears;Hypertelorism; joint hypermobility; long philtrum; shawl scrotum; short stature; Short/Broad Hands; widow's peak
			42 (I-1)	NR*	Cow's lick;Hypertelorism; Interphalangeal joint contractures; long philtrum; Short Stature; Widow's peak
<b>Altıncık et al. [31]</b>	2013	Turkey	43	7	Amblyopia; astigmatism; borderline mental retardation;Brachydactyly/wide fingers; broad NA†sal bridge; cryptorchidism; Frontal bossing; Hypertelorism; interdigital webbing; long philtrum; short stature; Short/Broad Hands
<b>Völter et al. [32]</b>	2014	Germany	44	9	ADHD; Broad NA†sal bridge; cleft lip palate;Clinodactyly; hemianopsia; hyperNA†sal speech; hypertelorism; interdigital webbing; long philtrum; low set ears; metatarsus varus; microcephaly; Midface hypoplasia; NA†sal ectopic teeth; ptosis; round face; short neck; Short stature; Short/Broad Hands; telecanthus; Von Willebrand disease
			45(Index)	16	Anteverted nostrils/Short Nose; Anteverted nostrils/Short Nose; hypertelorism; joint hypermobility; metatarsus varus; MicrogNA†thia; Myopathy; ogival palate; ptosis; shawl scrotum; short stature

<b>Al-Semari et al. [19]</b>	2013	Saudi Arabia	46(Bro1  )	14	Anteverted nostrils/Short Nose; Anteverted nostrils/Short Nose; hypertelorism; joint hypermobility; metatarsus varus; microgNA†thia; ogival palate; pectus excavatum; shawl scrotum; short stature
			47(Bro2  )	21	Anteverted nostrils/Short Nose; Anteverted nostrils/Short Nose; hypertelorism; joint hypermobility; metatarsus varus; microgNA†thia; ogival palate; shawl scrotum; short stature
			48(Nep¶1 )	1	Anteverted nostrils/Short Nose; Anteverted nostrils/Short Nose; hypertelorism; joint hypermobility; metatarsus varus; MicrogNA†thia; ogival palate; pectus excavatum; ptosis; shawl scrotum; short stature
			49(Nep¶2 )	4	Anteverted nostrils/Short Nose; Anteverted nostrils/Short Nose; coxa magNA†; hypertelorism; joint hypermobility; metatarsus varus; microgNA†thia; myopathy; ogival palate; shawl scrotum; short stature
<b>Niida et al.[33]</b>	2014	Japan	50	6	Anteverted nostrils/Short Nose; brachydactyly/wide fingers; clinodactyly; cryptorchidism; hypertelorism; interdigital webbing; ptosis; round face; shawl scrotum

Pérez-Coria et al. [4]	2015	Mexico	51(1)	6	Anteverted nostrils/Short Nose; bottom lip fold; brachydactyly/wide fingers; clinodactyly; cryptorchidism; dental malocclusion; dysplastic ears; frontal bossing; hypertelorism; interdigital webbing; joint hypermobility; long philtrum; midface hypoplasia; obesity; shawl scrotum; short stature; simian creases; wide feet; widow's peak
			52(4)	12	Bottom lip fold; brachydactyly/wide fingers; camptodactyly; Cryptorchidism; Dental malocclusion; downward slant palpebral fissures; dysplastic ears; hypertelorism; interdigital webbing; joint hypermobility; Long Philtrum; Midface Hypoplasia; Obesity; Psychomotor retardation; ptosis; shawl scrotum; Short stature; syndactyly; wide feet; widow's peak
			53(5)	4	Anteverted nostrils/Short Nose; bottom lip fold; brachydactyly/wide fingers; camptodactyly; clinodactyly; cryptorchidism; downward slant palpebral fissures; dysplastic ears; hypertelorism; inguiNA†I hernia; interdigital webbing; joint hypermobility; long philtrum; midface hypoplasia; ptosis; shawl scrotum; short stature; simian crease; umbilical hernia; wide feet; widow's peak

<b>GE et al. [20]</b>	2015	China†	54	2	Blepharophimosis; Bone age retarded; Brachydactyly/wide fingers; enlarged liver; groove below upper lip; hypertelorism; inguinal hernia; low set ears; short stature; splenic cyst; swollen testicles
<b>Pariltay et al. [34]</b>	2016	Turkey	55	14	Anteverted nostrils/Short Nose; Brachydactyly/wide fingers; broad nasal bridge; cryptorchidism; dysplastic ears; hypertelorism; large fontanelle/sutures; ogival palate; shawl scrotum; simian crease; widow's peak
<b>Griffin et al. [35]</b>	2016	USA§	56(IV-1)	1	Ankyloglossia; Anteverted nostrils/Short Nose; astigmatism; brachydactyly/wide fingers; clinodactyly; clubbed feet; cryptorchidism; diastema; downward slant palpebral fissures; dysplastic ears; interphalangeal joint contracture; low set ears; metatarsus varus; ptosis; round face; shawl scrotum; simian creases; still's murmur

Hamzeh et al. [36]	2017	UAE‡	57(IV-3)	7	ADHD;Anteverted nostrils/Short Nose; blepharophimosis; brachydactyly/wide fingers; broad NA†sal bridge; clinodactyly; Development delayed; downward slant palpebral fissures; dysplastic ears; epicanthal fold; frontal bossing; hypertelorism; interdigital webbing; Low set ears; microgNA†thia; ogival palate; pectus carina†tum; pectus excavatum; ptosis; short columella; short neck; short philtrum; Short stature; simian creases; sleep disorder; social/language skills delayed; syndactyly; thin upper lip vermillion; widely spaced nipples; widow's peak
			58(IV-5)	3	ADHD; Anteverted nostrils/Short Nose;Anteverted nostrils/Short Nose; Blepharophimosis; brachydactyly/wide fingers; Broad NA†sal bridge; clinodactyly; Development delayed; downward slant palpebral fissures; dysplastic ears; epicanthal fold; Frontal bossing; hypertelorism; long uvula; Neurocognitive development delayed; overactive frontalis muscle; poor bell's phenomenon; ptosis; short philtrum; short neck; short stature; Short/Broad Hands; telecanthus; widely spaced nipples; widow's peak

### Legend

\*NR: Not reported; †NA: Not applicable; ‡UAE: United Arab Emirates; §USA: Unite



**Log-Scott syndrome and assessment of genotype-phenotype correlation: a systematic review**  
 de Lima Oliveira, Eliene Magda de Assis, Michel Campos Ribeiro, Analina Furtado Valadão, A

List of all informations collected from each patient included

Type of mutation	Splicing Mutation	Amino Acid Change	Protein
Missense/Nonsense (1565G>A)	NA†	Arg522His	p.R522H
Missense/Nonsense (1565G>A)	NA†	Arg522His	p.R522H
Gross Deletion (Incl.ex.9-12)	NA†	NA†	NA†

Missense/Nonsense (1829G>A)	NA†	Arg610Gln	p.R610Q
Missense/Nonsense (1829G>A)	NA†	Arg610Gln	p.R610Q
Missense/Nonsense (1829G>A)	NA†	Arg610Gln	p.R610Q

Small Insertion (528insC)	NA†	NA†	NA†
Small Insertion (528insC)	NA†	NA†	NA†
Missense/Nonsense (614G>T)	NA†	Ser205Ile	p.S205I

Small Deletion (982delC)	NA†	NA†	p.(His328Thrfs*32)
Gross Deletions (944- 975del32)	NA†	NA†	NA†
Missense/Nonsense (1193 A>C)	NA†	Glu380Ala	p.E380A
Missense/Nonsense (1328 G>A)	NA†	Arg443His	p.R443H

Small Deletion (1316-1319del AGCT)	NA†	NA†	NA†
Small Deletion (2530delG)	NA†	NA†	p.(Val844Trpfs*19)
Missense/Nonsense (c.1223G>A)	NA†	Arg408Gln	p.R408Q

Small Deletion (c.2192delA)	NA†	NA†	p.(Lys731Argfs*132)
Small Deletion (c.2192delA)	NA†	NA†	p.(Lys731Argfs*132)
Small Deletion (c.2192delA)	NA†	NA†	p.(Lys731Argfs*132)
Missense/Nonsense (c.1327G>T)	NA†	Arg443Leu	p.R443L
Missense/Nonsense (c.2221G>T)	NA†	Glu741Term	p.E741*

Missense/Nonsense (c.1396A>G)	NA†	Met466Val	p.M466V
Small Insertions (c.944dupC)	NA†	NA†	p.(Ala316Cysfs*4)
Small Insertions (c.944dupC)	NA†	NA†	p.(Ala316Cysfs*4)

Gross Deletions (ENTIRE GENE)	NA†	NR*	NR*
Small Deletions (806delC)	NA†	NA†	p.(Ala269Valfs*91)
Missense/Nonsense (c.1205G>A)	NA†	Arg402Gln	p.R402Q
Missense/Nonsense (c.1590T>A)	NA†	Tyr530Term	p.Y530*



Small Deletions (1620delC)	NA†	NA†	Asp540Glufs*11
Missense/Nonsense (c.1673C>G)	NA†	Ser558Trp	p.S558W
Splicing (c.1935+3A>C)	IVS11 ds A-C +3	NA†	NA†
Missense/Nonsense (c.1966C>T)	NA†	Arg656Term	p.R656*

Missense/Nonsense (c.1966C>T)	NA†	Arg656Term	p.R656*
Missense/Nonsense (c.1966C>T)	NA†	Arg656Term	p.R656*
Small Deletions (2020_2020 del GAG)	NA†	NA†	Glu676del
Missense/Nonsense (c.2242A>G)	NA†	Lys748Glu	p.K748E

Missense/Nonsense (c.175C>T)	NA†	Gln59Term	p.Q59*
Missense/Nonsense (c.175C>T)	NA†	Gln59Term	p.Q59*
Duplication	NA†	NA†	NA†
Small deletions (c.2016-35delA)	NA†	NA†	NA†

Small deletions (c.2016-35delA)	NA†	NA†	NA†
Small deletions (c.2016-35delA)	NA†	NA†	NA†
Splicing (c.482-2A>G)	IVS2 as A-G -2	NA†	NA†
Missense/Nonsense (c.1468C>T)	NA†	Gln490Term	p.Q490*
Missense/Nonsense (c.1341G>A)	NA†	Trp447Term	p.W447*

Missense/Nonsense (c.1341G>A)	NA†	Trp447Term	p.W447*
Missense/Nonsense (c.1341G>A)	NA†	Trp447Term	p.W447*
Missense/Nonsense (c.1341G>A)	NA†	Trp447Term	p.W447*
Missense/Nonsense (c.1341G>A)	NA†	Trp447Term	p.W447*
Missense/Nonsense (c.1340G>A)	NA†	Trp447Term	p.W447*

Missense/Nonsense (c.1138G>T)	NA†	Glu380Term	p.E380*
Missense/Nonsense (c.1990C>T)	NA†	Gln664Term	p.Gln664*
Missense/Nonsense (c.1990C>T)	NA†	Gln664Term	p.Gln664*

Missense/Nonsense (c.1270A>G)	NA†	Asn424Asp	p.N424D
Splicing Mutations (c.1340+2T>A)	IVS6 ds T-A +2	NA†	NA†
Missense/Nonsense (c.2761C>T)	NA†	Gly757Arg	p.G757R

Small Deletions (c.53delC)	NA†	NA†	p.(Pro18Argfs*106)
Small Deletions (c.53delC)	NA†	NA†	p.(Pro18Argfs*106)

† = United States of America; ||bro: Brother; ¶nep= Nephew

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**tematic review**  
lfredo Orrico

Insertions/Duplications
NA†
NA†
NA†

NA†

NA†

NA†



NA†

NA†

NA†

NA†

NA†

NA†

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NA†

NA†

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NA†

NA†

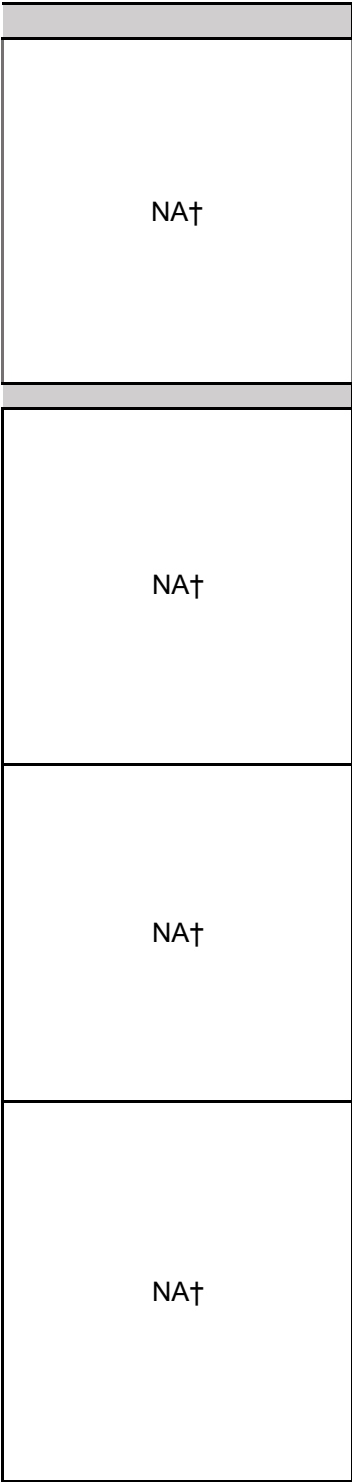
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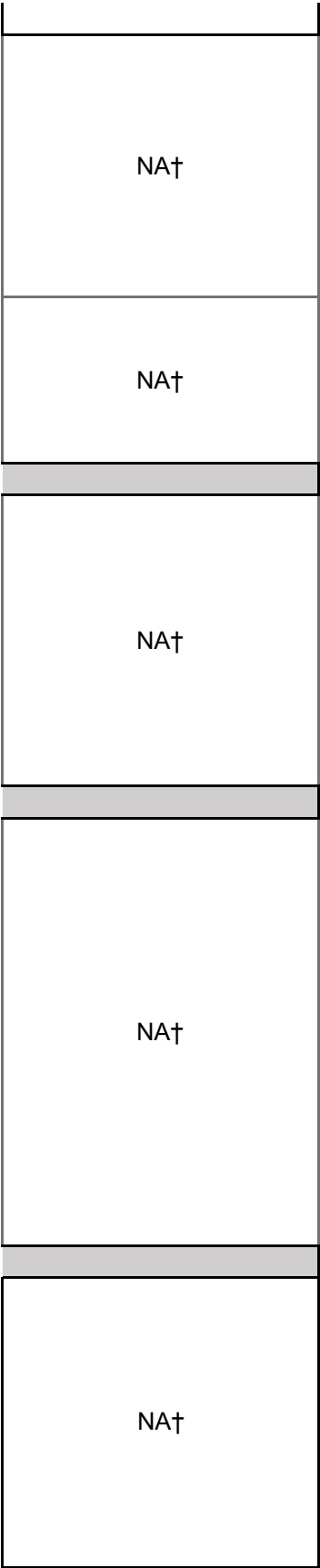
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NA†



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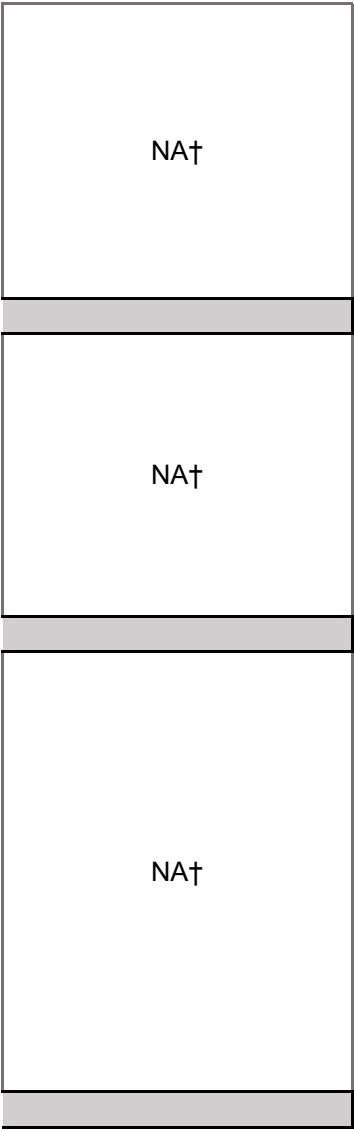
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