**Supplementary Table 1.** The clinical features associated with the genetic variants in DCAF17 reported in the literature.

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| --- | --- | --- | --- |
| **#** | **Genetic variant** | **Ref.** | **Clinical phenotype** |
| 1 | c.436delC | [1] | Progressive appearance of abnormal posturing and twisting movements, absence of secondary sexual characteristics, amenorrhea, diabetes mellitus, generalized dystonia, intellectual disability , edentulism, alopecia |
|  |  | [2] | Alopecia, intellectual disability, deafness, hypoplastic uterus, hypothyroidism, hyperlipidemia, diabetes mellitus along with end-stage renal disease, progressive gait deterioration, epilepsy, and T-wave abnormalities on ECG |
|  |  |  | Patient 1: Hypotrichosis, focal choreoathetoid, dystonic movements, progressive gait abnormalities, dysarthria, moderate intellectual impairment, cerebral palsy with extrapyramidal signs, delayed puberty, growth retardation, hypothyroidism, mild hypotrichosis, uncorrected visual acuity, and mild scoliosis  Patient 2: Frontotemporal hypotrichosis, intellectual disability, hypoplastic uterus, absence of both ovaries, uncorrected visual acuity and myopic fundus in both eyes  Patient 3: Frontotemporal hypotrichosis, intellectual disability, extrapyramidal abnormalities, delayed puberty, growth retardation, uncorrected visual acuity, and hypothyroidism |
|  |  |  | Family 1:  Patient 1: Delayed puberty, myopia and moderate sensorineural hearing loss, partial frontal alopecia, and borderline intellectual disability  Patient 2: Delayed puberty, primary amenorrhea, frontal alopecia, bilateral keratoconus, bilateral mild sensorineural deafness, borderline intellectual disability, and a hypoplastic uterus  Patient 3: Delayed puberty, primary amenorrhea, frontal alopecia, myopia, mild sensorineural hearing loss, borderline intellectual disability, and a hypoplastic uterus  Family 2:  Patient 4: Delayed puberty, primary amenorrhea, partial alopecia, borderline intellectual disability, and mild sensorineural deafness  Patient 5: Delayed puberty, hyperactivity, mild intellectual disability, and partial alopecia  Patient 6: Hyperactivity, mild intellectual disability, partial alopecia, and white matter hypomyelination  Patient 7: Hyperactivity, mild intellectual disability, and partial alopecia |
|  |  | [3] | Patient 1: Hypogonadotropic hypogonadism, lack of sexual development, alopecia, delayed primary and secondary tooth eruption, hyperopia, myopia with astigmatism, and microcephaly  Patient 2: Primary amenorrhea, hypoplastic ovaries and uterus |
|  |  | [4] | Hypogonadism, alopecia, diabetes mellitus, and different degrees of intellectual disability ranging from mild to severe |
|  |  | [5] | Hypogonadism, diabetes mellitus, alopecia, hypothyroidism, keratoconus, and deafness |
|  |  | [6] | Hypogonadism, alopecia, diabetes mellitus, intellectual disability, and extrapyramidal features |
|  |  | [7] | Patient 1: Mild extrapyramidal symptoms, primary amenorrhea, gonadal dysfunction, diabetes mellitus, alopecia, hypothyroidism, and lack of sexual development  Patient 2 and 3: Primary amenorrhea, alopecia, diabetes mellitus, and lack of sexual development |
|  |  | [8] | Dystonia (65.7%), intellectual disabilities (36.8%), sensorineural hearing loss (31.5%), and extrapyramidal rigidity (5.2%) |
|  |  | [9] | Patient 1: Dystonia, gait difficulties, mild cerebral atrophy, and decline in language and cognitive functions  Patient 2: Asymptomatic |
|  |  | [10] | sensorineural hearing loss and dystonia |
|  |  | [11] | Dystonia and abnormalities in pattern reversal visual EPs (PRVEPs), and somatosensory EPs (SSEPs). |
|  |  | [12] | Progressive gait difficulties and truncal dystonia |
| 2 | c.1A>G | [13] | Moderate/mild deafness, hair loss, intellectual disability and failure to develop secondary sexual characteristics |
| 3 | c.270delA | [14] | Ectodermal appendages, deafness, and failure to develop secondary sexual characteristics |
| 4 | c.321 + 1 G > A | [15] | Patient 1: Alopecia, complete edentulism, sensorineural deafness, moderate intellectual disability, primary amenorrhea, and failure to develop secondary sexual characteristics  Patient 2: Alopecia, partial edentulism, sensorineural deafness, moderate intellectual disability, blepharospasm, primary amenorrhea, and failure to develop secondary sexual characteristics  Patient 3 and 4: Alopecia, sensorineural deafness, moderate intellectual disability, and failure to develop secondary sexual characteristics  Patient 5: Alopecia, sensorineural deafness, moderate intellectual disability, dystonia, choreoathetosis, primary amenorrhea, and failure to develop secondary sexual characteristics  Patient 6: Alopecia, sensorineural deafness, moderate intellectual disability, dystonia, choreoathetosis, spastic quadriplegia, dysarthria, dysphagia, and failure to develop secondary sexual characteristics |
| 5 | c.1091+1G>A | [16] | Patient 1: Slowness of movements, dysarthria, dysphagia, mild intellectual disability, alopecia, hypogonadotropic hypogonadism, diabetes mellitus, hypophonic speech and hypokinesia |
|  |  | [17] | Patient 1:  Slowness of movements, dysarthria, dysphagia, mild intellectual disability, alopecia, hypogonadotropic hypogonadism, diabetes mellitus, hypophonic speech, hypokinesia AND iron deposition in the brain |
| 6 | c.270 dup | [18] | Patient 1: Primary amenorrhea, delayed puberty, learning disabilities, mild intellectual disability, hyper-hypogonadotropic hypogonadism, prepubertal internal genitalia, non-immune-insulinopenic diabetes mellitus, secondary hypothyroidism, sensorineural deafness, and dysarthria |
| 7 | c.127-3delTAGinsAA | [19] | Hypothyroidism, frontotemporal alopecia, dystonia, hypogonadism, and intellectual disability |
| 8 | c.1423‐1\_1425delGACA | [20] | Alopecia, intellectual disability, hypogonadism, mild sensory neural deafness, facial dysmorphism, delayed speech, language development, and extrapyramidal features |
| 9 | c.1091+2T>C | [21] | Dystonia, mild intellectual disability, hypergonadotrophic hypogonadism, primary amenorrhea, diabetes mellitus, hypothyroidism, bilateral sensorineural hearing loss and alopecia |
| 10 | c.1488\_1489delAG | [22] | Alopecia, hypogonadism, hypothyroidism, intellectual disability, anemia and thrombocytopenia |
| 11 | c.1111delA | [23] | Alopecia, diabetes mellitus, intellectual disability, hypothyroidism and hypogonadism |
| 12 | c.906 G>A | [19] | Patient 1: Alopecia, diabetes mellitus, hypogonadism, intellectual disability, deafness, extrapyramidal signs and myocardial infarction  Patient 2: Alopecia, hypogonadism, intellectual disability and myocardial infarction  Patient 3: Alopecia, diabetes mellitus, hypogonadism, intellectual disability, deafness, extrapyramidal signs and amenorrhea |
| 13 | c.1238delA  c.459- 7\_499del | [24] | Patient 1,2: Amenorrhea, alopecia, sensorineural hearing loss, diabetes mellitus and intellectual disability  Patient 3: Alopecia, sensorineural hearing loss, diabetes mellitus and intellectual disability |

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