ORIGINAL ARTICLE

Characteristic facial features and cortical blindness distinguish the DOCK7-related epileptic encephalopathy

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Abstract

**Background:** The epileptic encephalopathies display extensive locus and allelic heterogeneity. Biallelic truncating DOCK7 variants were recently reported in five children with early-onset epilepsy, intellectual disability, and cortical blindness, indicating that DOCK7 deficiency causes a specific type of epileptic encephalopathy.

**Methods:** We identified 23- and 27-year-old siblings with the clinical pattern reported for DOCK7 deficiency, and conducted genome-wide linkage analysis and WES. The consequences of a DOCK7 variant were analyzed on the transcript and protein level in patients’ fibroblasts.

**Results:** We identified a novel homozygous DOCK7 frameshift variant, an intragenic tandem duplication of 124-kb, previously missed by CGH array, in adult patients. Patients display atrophy in the occipital lobe and pontine hypoplasia with marked pontobulbar sulcus, and focal atrophy of occasional cerebellar folia is a novel finding. Recognizable dysmorphic features include normo-brachycephaly, narrow forehead, low anterior and posterior hairlines, prominent ears, full cheeks, and long eyelashes. Our patients function on the level of 4-year-old children, never showed signs of regression, and seizures are largely controlled with multi-pharmacotherapy. Studies of patients’ fibroblasts showed nonsense-mediated RNA decay and lack of DOCK7 protein.

**Conclusion:** DOCK7 deficiency causes a definable clinical entity, a recognizable type of epileptic encephalopathy.

**KEYWORDS**
cortical blindness, DOCK7, epileptic encephalopathy, nonsense-mediated RNA decay, recognizable syndrome

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1 | INTRODUCTION

The early infantile epileptic encephalopathies (EIEE) represent a large and genetically heterogeneous group of neurodevelopmental disorders diagnosed during early childhood. Variable degrees of social, cognitive, motor, language, and behavioral impairments are observed. Seizures supposedly contribute to developmental impairment and regression (Scheffer et al., 2017). Whole exome sequencing (WES) or targeted gene panels enable a genetic diagnosis in about 30% of EIEE patients. Genetic diagnoses can lead to more accurate treatment in up to 25% of the epilepsy patients, that is, they are required for precision medicine (Heyne et al., 2019; Moller et al., 2019). However, genes whose mutations are known to cause EIEE are most often associated with overlapping and non-specific phenotypes, complicating the establishment of etiological diagnoses.

Recently, a specific phenotype of EIEE (EIEE23, OMIM 615859) was suggested based on the findings in five children aged 3–10 years, who harbored biallelic truncating mutations in the DOCK7 (dedicator of cytokinesis 7) gene (OMIM 615730), that were supposed but not shown to trigger nonsense-mediated mRNA decay (NMD) (Bai et al., 2019; Perrault et al., 2014; Turkdogan et al., 2019). DOCK7 encodes a guanine nucleotide exchange factor (GEF) that plays a role in axon formation and neuronal polarization (Watabe-Uchida et al., 2006).

We report an adult sibling-pair displaying the pattern of EIEE23; these patients function on the level of 4-year-old children with largely controlled seizures. We identify a novel homozygous truncating DOCK7 variant, and show that it abrogates protein production.

2 | MATERIALS AND METHODS

2.1 | Ethical compliance

Written informed consent for molecular genetic studies and publication of data was obtained, and the ethics committee of the Medical University of Innsbruck approved the study. Linkage analysis and WES were performed as reported (Baumann et al., 2017; Waich et al., 2020), and breakpoint sequencing and functional studies are described in Supporting Information.

3 | RESULTS

3.1 | Clinical characteristics

The two female patients (P1 and P2) are 27 and 23 years old. They were born at term after unremarkable pregnancy and delivery to healthy, consanguineous Austrian parents. Their neonatal course was uneventful, except for the discovery of an atrial septal defect in P2, for which the child was operated on at 5 years of age. Both sisters presented with infantile spasms at 6 months of age, which occurred as many as 50 times a day. Over the next months, both patients showed different types of seizures, including myoclonus, partial complex seizures with rotation of the head, drop attacks, and tonic seizures. Control was initially poor in both sisters despite the administration of multiple antiepileptic drugs in various combinations. Electroencephalography (EEG) performed at 11 months of age in P1 and at 5 months of age in P2 showed a pattern consistent with hypsarrhythmia. Subsequent EEG studies showed multifocal epileptic activity in both sisters. Current antiepileptic therapy consists of levetiracetam, clobazam, zonisamide, and midazolam to terminate prolonged seizures.

Lack of reaction to visual stimuli was evident during the first months of life; a fine horizontal and vertical nystagmus in both eyes and lack of object fixation first suggested Leber congenital amaurosis (LCA) in both patients. However, pupillary reactions and fundoscopy were normal in both sisters, and the scotopic flash evoked visual potentials (FEVP) showed a normal waveform of markedly decreased amplitude in both patients, which led to a diagnosis of cortical blindness. A photopic Ganzfeld electroretinogram (GF-ERG) showed mildly reduced amplitudes in P1 at 20 years of age, indicating some degree of retinal dysfunction; scotopic testing was not possible due to lack of cooperation. Currently, both individuals still display grossly abnormal visual pursuit, but can ambulate with moderate speed in known environment. P1 correctly identifies large objects and colors, and P2 has nearly no vision.

Patients’ adult body height and occipitofrontal head circumference measures are 163, 164, 56 and 54 cm, respectively, and they are mildly obese. Patients show recognizable facial features (Figure 1, P1 and P2). Both patients entered and completed puberty at appropriate age.

P1 and P2 started to walk at 20 months of age, and today patients require assistance to ambulate in unknown environment. Both patients can eat by themselves, can brush their teeth unaided, and are continent by day since age 12 years. Both patients rarely point or use their hands to communicate, but can use objects and perform easy tasks in a sheltered workshop; P1 can assemble a 48-piece puzzle, and P2 can cut vegetables in the community kitchen. Both patients speak grammatically correct sentences, referring to simple subjects, and understand simple commands, and can designate body parts on demand. They can smile in and out of social context. They display nearly no visual contact. Overall, their skills were rated as on the level of 4-year-old children by using the SON-R 2½-7 non-verbal intelligence test, and by non-formal clinical assessment, and they never showed...
signs of regression. Neurological examination was otherwise unremarkable.

A metabolic work-up, including plasma triglycerides, cholesterol, amino acid concentrations, and urine organic acid chromatography, was normal in both sisters. All previously reported structural brain abnormalities were present in both siblings, but the focal atrophy of cerebellar folia is a novel finding (Figure 1a–g).

Major clinical findings of our and of reported patients are compiled in Table 1.

### 3.2 Genetic and protein studies

Linkage analysis with all individuals in generations III-V excluded 97% of the genome to harbor the disease locus (Figure 2a,b). WES analysis in P1 identified homozygosity for an intragenic 124-kb tandem duplication in \textit{DOCK7} (NC_000001.11:g.62527474_62651054dup, NM_001271999.1:c.390_3936dup), comprising exons 5–31, as determined by genomic breakpoint and by fibroblast cDNA sequencing (Figure 2c,d). The duplication causes a frameshift and DOCK7 deficiency (Figure 2e) via NMD, as demonstrated by partial mRNA rescue with NMD inhibitor puromycin prior to fibroblast culture harvesting. To detect residual DOCK7 protein in whole-cell lysates, a polyclonal antibody raised in Rabbit with a synthetic peptide corresponding to a region within amino acids 175–225 of human DOCK7 was used.

This duplication also contains the complete \textit{ANGPTL3} gene, which resides within exon 14 of \textit{DOCK7} and is transcribed from the other strand. \textit{ANGPTL3} deficiency
| Reference         | This study | Bai et al. (2019) | Perrault et al. (2014) | Patient A-1 | Patient A-2 | Patient B-1 | Turkdoğan et al. (2019) |
|-------------------|------------|-------------------|------------------------|-------------|-------------|-------------|-------------------------|
| Patient ID        | P1         | P2                | C                      | Patient A-1 | Patient A-2 | Patient B-1 | D                       |
| Age (years)       | 27         | 23                | 3                      | 7           | 5           | 10          | 3                       |
| Sex               | Female     | Female            | Female                 | Female      | Female      | Male        |                         |
| Ethnicity         | Austrian   | Austrian          | Chinese                | French-Canadian | French-Canadian | French      | Turkish                 |
| Parental consanguinity | Yes   | Yes               | No                     | No          | No          | No          | Yes                     |
| DOCK7 variants    | Homozygous | Homozygous        | Compound-het.          | Compound-het. | Compound-het. | Compound-het. | Homozygous              |
| Variant type      | Intragenic 124 kb tandem duplication | Intragenic 124 kb tandem duplication | Splice/stop | Stop/frameshift | Stop/frameshift | Stop/frameshift | Stop                     |
| cDNA variant      | c.390_3936dup | c.390_3936dup | c.390_3936dup | c.390_3936dup | c.390_3936dup | c.390_3936dup | c.390_3936dup           |
| Protein variant   | Loss of protein | Loss of protein | Loss of protein | Loss of protein | Loss of protein | Loss of protein | Loss of protein         |
|蛋白位置 | NC_000001.10:g.6299134 _63116725dup | NC_000001.10:g.6299134 _63116725dup | NC_000001.10:g.6299134 _63116725dup | NC_000001.10:g.6299134 _63116725dup | NC_000001.10:g.6299134 _63116725dup | NC_000001.10:g.6299134 _63116725dup | NC_000001.10:g.6299134 _63116725dup |
| cMRI | (at age 20 years) Abnormally marked pontobulbar sulcus, mild pontine hypoplasia, atrophy in occipital white and gray matter | (at age 16 years) Abnormally marked pontobulbar sulcus, mild pontine hypoplasia, atrophy in occipital white and gray matter | (at age 3 years) Abnormally marked pontobulbar sulcus, mild pontine hypoplasia, atrophy in occipital white and gray matter | (at age 25 months) Abnormally marked pontobulbar sulcus, mild pontine hypoplasia, atrophy in occipital white and gray matter | (at age 8 months) Mild hypoplasia of the corpus callosum | (at age 2 years) Abnormally marked pontobulbar sulcus, mild pontine hypoplasia, atrophy in occipital white and gray matter | (at age 33 months) Marked pontobulbar sulcus, pontine hypoplasia, atrophy in occipital white and gray matter |
| EEG, initial and on follow-up | Hypsarrhythmia, multifocal epileptic activity | Hypsarrhythmia, multifocal epileptic activity | Hypsarrhythmia, multifocal epileptic activity | Hypsarrhythmia, multifocal epileptic activity | Hypsarrhythmia, multifocal epileptic activity | Hypsarrhythmia, multifocal epileptic activity | Hypsarrhythmia, multifocal epileptic activity |
| Eye revulsion, rhythmic arm and body movements. Short absences | Eye revulsion, rhythmic arm and body movements. Short absences | Eye revulsion, rhythmic arm and body movements. Short absences | Eye revulsion, rhythmic arm and body movements. Short absences | Eye revulsion, rhythmic arm and body movements. Short absences | Eye revulsion, rhythmic arm and body movements. Short absences | Eye revulsion, rhythmic arm and body movements. Short absences | Eye revulsion, rhythmic arm and body movements. Short absences |
| Status epilepticus frequent focal motor seizures | Status epilepticus frequent focal motor seizures | Status epilepticus frequent focal motor seizures | Status epilepticus frequent focal motor seizures | Status epilepticus frequent focal motor seizures | Status epilepticus frequent focal motor seizures | Status epilepticus frequent focal motor seizures | Status epilepticus frequent focal motor seizures |
| Multifocal epileptic activity with occasional electroclinical spasms | Multifocal epileptic activity with occasional electroclinical spasms | Multifocal epileptic activity with occasional electroclinical spasms | Multifocal epileptic activity with occasional electroclinical spasms | Multifocal epileptic activity with occasional electroclinical spasms | Multifocal epileptic activity with occasional electroclinical spasms | Multifocal epileptic activity with occasional electroclinical spasms | Multifocal epileptic activity with occasional electroclinical spasms |

(Continues)
**Table 1 Continued**

| Facial features | Low anterior and posterior hairline, highly arched palate, some periorbital fullness, telecanthus, long eyelashes, a broad nasal tip. Low-set and protruding ears. Smooth and short philtrum and thin upper lip | Low anterior and posterior hairline, highly arched palate, gingival maldevelopment, telecanthus, long eyelashes, low-set, abnormally shaped and protruding ears, periorbital fullness, broad nasal tip, large root | Low posterior hairline, some periorbital fullness, telecanthus, long eyelashes, a broad nasal tip with anteverted nares | Low anterior hairline, some periorbital fullness, telecanthus, long eyelashes, a broad nasal tip with anteverted nares | Low anterior hairline, thick eyebrows, synophrys, telecanthus, long eyelashes, enophthalmia, large and prominent nasal root, a bulbous nasal tip, thick helix and earlobes, a short philtrum, full lips and everted lower lip, spaced incisors |
|-----------------|-------------------------------------------------|-------------------------------------------------------------------------------------------------|-----------------------------------------------------------------|-----------------------------------------------------------------|-----------------------------------------------------------------|
| Eye abnormality | Lack of reaction to visual stimuli during the first months of life; a fine horizontal and vertical nystagmus in both eyes, and lack of object fixation. Normal pupillary reactions and fundoscopy. FEVP: normal waveform of markedly decreased amplitude, cortical blindness. Nearly no vision | Lack of ocular reaction to visual stimuli. Binocular optometric obstacles. Horizontal optokinetic nystagmus, left strabismus. Flash evoked visual potentials (FEVP) show abnormal waveform including a longer latency in right eye and decreased amplitude in both eyes cortical blindness | Normal eye movements, pupillary reaction, and fundus. Lack of ocular reaction to visual stimulus, binocular optometric obstacles, cortical blindness. Can follow a moving object, but does not see well enough to play with toys | Normal eye movements, pupillary reaction, and fundus. Lack of ocular reaction to visual stimulus, binocular optometric obstacles, cortical blindness. Can follow a moving object, but does not see well enough to play with toys | Lack of ocular reaction to visual stimulus, cortical blindness. Evoked visual potentials were unremarkable, but electroretinographic (ERG) traces were ambiguous. Ophthalmological examinations repeated at 2 and 9 years of age showed unchanged retinal aspect and normal ERG traces, leading to the diagnosis of cortical blindness. Currently, there are wandering eye movements and a complete absence of reaction to visual threat and light stimulation. |
| Heart           | Normal | Atrial septal defect | Atrial septal defect | Aortic supravalvular stenosis, bicuspid valve | Normal |
| Language        | Understands and speaks grammatically correct sentences of a few words, make use of cell phones, follow simple commands, can designate body parts on demand | Lack of speech | Lack of speech, understands a few simple commands | Speaks 30 words and associates two words. She can designate body parts on demand and understand simple commands | Repeating three words, understands simple commands |

(Continues)
was associated with a form of hypobetalipoproteinemia (Musunuru et al., 2010); our patients apparently have four intact copies of ANGPTL3 and repeatedly normal serum lipid levels.

Conventional karyotyping after GTG-banding at a 500 band resolution showed normal female karyotypes, 46,XX. Chromosomal microarray analysis (Illumina HumanCytoSNP-12v2 BeadChip SNP array with 300 k markers) in P1 had missed the homozygous DOCK7 duplication due to a sparsity of markers in the region.

We report here, to the best of our knowledge, for the first time the outcome of DOCK7 deficiency in two adult patients and corroborate the hypothesis that there is a distinctive EIEE23 phenotype that consists of an infantile-onset epilepsy, severe neurodevelopmental delay, cortical blindness, and the typical facial features and common brain abnormalities described above. Additional developmental brain abnormalities described in single patients each, such as focal atrophy of cerebellar folia in P2, the interdigitation of gyri across the interhemispheric fissure and the absence of the interventricular septum (Turkdogan et al., 2019), and pachygyria and dilation of lateral ventricles (Bai et al., 2019).

In six of seven patients reported to date, epilepsy was largely controlled by multi-pharmacotherapy, onward from the ages of 16 months to 6 years. Importantly, our two patients continuously acquired skills, in particular attention span and social communication skills over the years, and participate in daily life at home and outside. This outcome appears encouraging with respect to the five children previously described with biallelic truncating DOCK7 mutations, at ages 3–10 years, and who all showed moderate hypotonia, walking at 22 months, can walk without help in known environments. Can grasp objects, but not point or communicate with hands. Brings a spoon to her mouth, but cannot eat by herself at 35 months delayed gross and fine motor functions, lack of any visual contact with faces or objects.

TABLE 1 Continued

| Psychomotor development | Started to walk at 20 months of age. Today, requires assistance to ambulate in unknown environment. Can eat by herself, can brush her teeth unaided, and is continent by day since age 12 years. Rarely point or uses her hands to communicate, but can use objects and perform easy tasks in a sheltered workshop. Displays nearly no visual contact. | Walking at 20 months of age. Today, requires assistance to ambulate in unknown environment. Can eat by herself, can brush her teeth unaided, and is continent by day since age 12 years. Rarely point or uses her hands to communicate, but can use objects and perform easy tasks in a sheltered workshop. Displays nearly no visual contact. | Walking at 20 months, with help. Cannot eat by herself, does not point or use her hands to communicate. | Walking at 28 months, can run, cannot jump. Eats with a spoon and has a pincer grasp but cannot point to objects. | At 35 months delayed gross and fine motor functions - about at the developmental level of 15 and 8 months, respectively, lack of any visual contact with faces or objects. |

aNone of these variants is listed in gnomAD.
bCoding sequence nomenclature refers to NCBI reference NM_001271999.1/Ensembl reference ENST00000454575.6.
cLoss of protein as determined in patients' fibroblasts; FEVP, Scotopic Flash evoked visual potentials; GF-ERG, Ganzfeld electroretinogram.

4 | DISCUSSION

This table lists the details of each patient's development. The table includes information on walking, eating, and other developmental milestones. It also notes any specific medical conditions or treatments that have been observed, such as seizures and visual impairments. The table is structured in a clear and concise manner, making it easy to understand and compare the different patients.

Psychomotor development

| Patient | Walking at 20 months of age | Walking at 20 months, with help | Walking at 28 months, can run | At 35 months delayed gross and fine motor functions |
|---------|-----------------------------|--------------------------------|-----------------------------|---------------------------------------------------|
| P1      | Can eat by herself, can brush her teeth unaided, and is continent by day since age 12 years. Rarely point or uses her hands to communicate. | Cannot eat by herself, does not point or use her hands to communicate. | Can run, cannot jump. Eats with a spoon and has a pincer grasp but cannot point to objects. | About at the developmental level of 15 and 8 months, respectively, lack of any visual contact with faces or objects. |

This table provides a comprehensive overview of each patient's development, highlighting the progression of their abilities over time.
is sufficient to cause defects in neurogenesis (Nakamuta et al., 2017). DOCK7 is expressed in GABAergic interneurons in the central nervous system. The reduced ERG amplitudes in our patient might indicate the involvement of GABAergic retinal amacrine cells as well as DOCK7 expression in the retina, which would need to be addressed with further studies.

It is of interest that a strong conserved craniofacial-specific enhancer (identifier: GH01J062686) localizes within and around exon 1 of DOCK7 (Wilderman et al., 2018). It remains highly speculative to suggest that premature stop codons in the downstream coding sequence alter the pattern of transcription factor binding to this craniofacial-specific enhancer and thereby lead to the recognizable syndromic features of patients with DOCK7 deficiency.

Altogether, our observation validates the hypothesis that loss of DOCK7 function causes a recognizable form of EIEE, with the hallmarks of cortical blindness and common developmental brain abnormalities, and might potentially be clinically diagnosed based on the shared facial features.

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CONFLICT OF INTEREST
The authors declare that they have no competing interests.

AUTHOR CONTRIBUTIONS
EH and ARJ participated in the conception of the study. ARJ drafted the manuscript. All authors collected and analyzed data, interpreted the results, and revised the manuscript. All authors read and approved the final manuscript.

DATA AVAILABILITY STATEMENT
The data that support the findings of this study are available from the corresponding author upon reasonable request.
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SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section.

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