Ataxia-pancytopenia syndrome with \textit{SAMD9L} mutations

\textbf{ABSTRACT}

\textbf{Objective:} We describe the neurologic, neuroradiologic, and ophthalmologic phenotype of 1 Swedish and 1 Finnish family with autosomal dominant ataxia-pancytopenia (ATXPC) syndrome and \textit{SAMD9L} mutations.

\textbf{Methods:} Members of these families with germline \textit{SAMD9L} \textit{c.2956C>T, p.Arg986Cys, or c.2672T>C, p.Ile891Thr} mutations underwent structured interviews and neurologic and ophthalmologic examinations. Neuroimaging was performed, and medical records were reviewed. Previous publications on \textit{SAMD9L}-ATXPC were reviewed.

\textbf{Results:} Twelve individuals in both families were affected clinically. All mutation carriers examined had balance impairment, although severity was very variable. All but 1 had nystagmus, and all but 1 had pyramidal tract signs. Neurologic features were generally present from childhood on and progressed slowly. Two adult patients, who experienced increasing clumsiness, glare, and difficulties with gaze fixation, had paracentral retinal dysfunction verified by multifocal electroretinography. Brain MRI showed early, marked cerebellar atrophy in most carriers and variable cerebral periventricular white matter T2 hyperintensities. Two children were treated with hematopoietic stem cell transplantation for hematologic malignancies, and the neurologic symptoms of one of these worsened after treatment. Three affected individuals had attention deficit hyperactivity disorder or cognitive problems. Retinal dysfunction was not previously reported in individuals with ATXPC.

\textbf{Conclusions:} The neurologic phenotype of this syndrome is defined by balance or gait impairment, nystagmus, hyperreflexia in the lower limbs and, frequently, marked cerebellar atrophy. Paracentral retinal dysfunction may contribute to glare, reading problems, and clumsiness. Timely diagnosis of ATXPC is important to address the risk for severe hemorrhage, infection, and hematologic malignancies inherent in this syndrome; regular hematologic follow-up might be beneficial. \textit{Neurol Genet} 2017;3:e183; doi: 10.1212/NXG.0000000000000183

\textbf{GLOSSARY}

ADHD = attention deficit hyperactivity disorder; ATXPC = ataxia-pancytopenia; mfERG = multifocal electroretinography; HSCT = hematopoietic stem cell transplantation.

Ataxia-pancytopenia (ATXPC; MIM 159550) syndrome is an autosomal dominant disease with early-onset gait and balance impairment, nystagmus, mild pyramidal signs, and marked cerebellar atrophy. Hematological abnormalities in ATXPC include pancytopenia, which may remain subclinical but can cause severe infections or hemorrhages. Through different genetic mechanisms in blood cells, the hematopoietic phenotype may be reverted. Elimination of the germline \textit{SAMD9L} mutation by loss of chromosome 7(q) can result in myeloid malignancies.\textsuperscript{1}
ATXPC has so far been described in 5 families, including the Swedish family (F1) and Finnish family (F2) reported here. Recently, missense mutations in sterile alpha motif domain–containing protein 9-like (SAMD9L) have been identified as the cause of ATXPC in 4 of these families. Following the genetic discovery, we studied the neurologic phenotype in F1 and F2.

**METHODS** Affected and unaffected family members of both families were examined by a neurologist. A structured interview and examination were performed including all clinical features previously described in ATXPC families and the Scale for the assessment and rating of ataxia. Neuroradiologic images were compiled. Eye movement examination was filmed and reviewed by a neuro-otologist. Medical records were obtained whenever possible. Results of previous blood cell counts were reviewed, and new blood samples were taken. DNA was extracted using standard methods from the peripheral blood or buccal swabs. One of the genetic analyses and clinical (hematologic) information have previously been published. For this study, F1 was expanded; newly included family members were tested with a custom droplet digital PCR TaqMan genotyping assay for the SAMD9L c.2956C>T mutation (primers available on request). Two patients underwent detailed ophthalmologic examination, including optical coherence tomography, full-field electroretinography, and multifocal electroretinography (miERG; appendix e-1 at Neurology.org/ng).

**RESULTS** Data were compiled from 21 members of F1 and 6 members of F2. The pedigrees, individual neurologic phenotype, and SAMD9L genotype are shown in figure 1 and table, and patient descriptions in appendix e-1 and videos 1–3. DNA was obtained from 18 members of F1 and 6 members from F2. All mutation carriers who were examined within this study showed neurologic signs except the 4-year-old mutation carrier, F1: V-2, who had no obvious neurologic signs or symptoms according to her parents. Individual F1: IV-5 declined participation. Thirteen family members were examined who were subsequently shown not to carry SAMD9L mutations; these did not have neurologic signs or symptoms.

F1: III-4 presented at our neurology clinic at age 53 years because of mild balance problems and a positive family history (figure 1). No SAMD9L mutation
| ID: ref | SAMD9L mutation(s) | AE/AD | Balance | Dysm | Dysart | Nystag | Strab | Retina | Pyram | Sens | Cog/beh | Cytopenia | MD |
|--------|---------------------|-------|---------|------|--------|--------|-------|--------|------|-----|---------|-----------|----|
| F1: I-2 | ND                  | 80*   | ++ <55  | NA   | NA     | NA     | NA    | NA     | NA   | NA | –       | –         |    |
| F1: II-3 | ND                  | 87*   | + + +53 | +    | +     | GEN    | NA    | +    | NA   | NA | –       | –         |    |
| F1: III-4 | c.2956C>T, p.Arg986Cys (nonhematopoietic cells/buccal swabs only) | 64    | + +15   | +    | +    | GEN   | –     | PRD   | AC   | Bab, LEHR | Vibr | –     | P; Neu |    |
| F1: III-5 | c.2956C>T, p.Arg986Cys | 58    | +       | NA   | NA   | NA     | +     | NA    | NA   | NA | –       | R, P, L; Inf | MDS (56) |
| F1: IV-3 | c.2956C>T, p.Arg986Cys; c.689C>A, p.Thr233Asn | 38    | +       | –    | –    | DBN   | +     | –    | LEHR | –   | –       | –         |    |
| F1: IV-4 | c.2956C>T, p.Arg986Cys; c.689C>A, p.Thr233Asn | 33    | +       | –    | –    | GEN   | DBN   | –    | PRD   | LEHR | –   | –       | –         |    |
| F1: IV-5 | c.2956C>T, p.Arg986Cys | 28    | NA      | NA   | NA | NA     | NA    | NA    | –    | Rr, TP, TL | –       |    |
| F1: V-1 | c.2956C>T, p.Arg986Cys | 6     | ++ (4*) | +    | –    | –     | V     | –    | –    | LEHR, S, Bab, AC | –   | –  | R, P, L | MDS (4) |
| F1: V-2 | c.2956C>T, p.Arg986Cys | 4     | –       | NA   | NA   | NA     | NA    | NA    | NA   | NA | –       | –         |    |
| F2: I-1 | c.2672T>C, p.Ile891Thr | 34    | + [32]  | +    | – | GEN   | DBN   | –    | NA   | LEHR, LLW | –   | LI, MI, ADHD susp | TP; Inf |    |
| F2: II-1 | c.2672T>C, p.Ile891Thr | 14    | + (5)   | –    | –    | –    | –    | NA    | –    | –   | ADHD | –         | Rr; Inf |    |
| F2: II-4 | c.2672T>C, p.Ile891Thr | 9     | + + (7) | +    | –    | H     | –    | NA    | LEHR, LLW, ppv | –   | ADHD (4) | R, P, L; Inf | MDS (1,5) |
| I-1<sup>2,3,5</sup> | c.3587G>C, p.Cys1196Ser | 54    | ++++adol | +    | +    | H     | +    | NR    | AC   | Bab | Vibr | –         | NR       |    |
| II-1<sup>2,3,5</sup> | c.3587G>C, p.Cys1196Ser | 10*   | ++ child | NR   | NR   | NR    | +    | NR    | NR   | NR | –       | Neu       | AML (10) |    |
| II-2<sup>2,3,5</sup> | c.3587G>C, p.Cys1196Ser | 5*    | ++ child | NR   | NR   | NR    | +    | NR    | NR   | NR | –       | R, P, L, Hem, Inf | –       |    |
| II-3<sup>2,3,5</sup> | c.3587G>C, p.Cys1196Ser | 9*    | ++      | NR   | NR   | V + H | +    | NR    | NR   | NR | –       | R, Neu, Inf | –       |    |
| II-4<sup>2,3,5</sup> | c.3587G>C, p.Cys1196Ser | 54    | ++++adol | +    | +    | V + H | NR   | NR    | Bab  | Vib | –       | R         | –       |    |
| II-5<sup>2,3,5</sup> | c.3587G>C, p.Cys1196Ser | 7*    | ++ child | NR   | V    | +    | NR    | NR    | NR   | –   | R, P, L | AMMoL (7) | –       |    |
| UW-AP II-3<sup>5</sup> | c.2640C>A, p.His880Gln | 79a   | ++ +5 to 50 | NR   | NR   | NR    | NR    | NR    | NR   | NR | –       | dem 69 | Hem | –       |
| UW-AP II-4<sup>5</sup> | c.2640C>A, p.His880Gln | 85*   | + +62   | +    | +    | V + H | NR   | NR    | NR   | Bab, LEHR | NR | –   | P | –       |
| UW-AP III-3<sup>5</sup> | c.2640C>A, p.His880Gln | 60    | + 30    | +    | +    | H > V | NR   | NR    | NR   | AC, LEHR | NR   | –   | R, P, L | –       |
| UW-AP III-5<sup>5</sup> | c.2640C>A, p.His880Gln | 16*   | – 16    | NR   | NR   | NR    | NR    | NR    | NR   | NR | –       | R, P, L; Hem | –       |    |
| UW-AP III-6<sup>5</sup> | c.2640C>A, p.His880Gln | 55    | ++ +25  | +    | +    | V + H | NR   | NR    | NR   | AC, LEHR | NR   | –   | NR       | –       |
| UW-AP III-8<sup>5</sup> | c.2640C>A, p.His880Gln | 50    | +25     | +    | NR   | V + H | NR   | NR    | NR   | AC, Bab, LEHR | NR | –   | NR       | –       |
| UW-AP IV-1<sup>5</sup> | c.2640C>A, p.His880Gln | 32    | – 18    | +    | –    | V + H | NR   | NR    | NR   | AC, LEHR | NR   | –   | –       | –       |
| UW-AP IV-2<sup>5</sup> | c.2640C>A, p.His880Gln | 30    | – 16    | +    | –    | V + H | NR   | NR    | NR   | –   | –       | –         | R, P, L | –       |
| UW-AP IV-3<sup>5</sup> | c.2640C>A, p.His880Gln | 25    | – 11    | +    | –    | H     | NR   | NR    | NR   | AC, LEHR | NR   | –   | R, P, L | –       |

Continued
| ID: ref | SAMD9L mutation(s)  | AE/AD | Balance | Dysm | Dysart | Retina | Pyram | Sens | Cog/beh | Cytopenia | MD |
|--------|-----------------------|-------|---------|------|--------|--------|-------|------|---------|-----------|----|
| NE 5   | -                     | 11    | 3       | 1    | NR     | V      | 1     | H    | NR      | NR        | Bab, LLW sPNP |
| NE 12  | +                     | 11    | 7       | 1    | V      | 1     | H    | NR   | NR      | Bab, LLW sPNP |
| NE 34  | ++                    | 111   | 111     | 5    | V      | 1     | H    | NR   | NR      | Bab, LLW sPNP |

**Abbreviations:**
- none; 15 11 5
- moderate; 15 11 5
- severe; AC
- ankle clonus; ADHD
- attention deficit hyperactivity disorder
- acute myelogenous leukemia; AMMoL
- acute myelomonocytic leukemia; ATXPC
- ataxia-pancytopenia; Bab
- balance; LWD
- lower extremity hyperreflexia; LI
- normochromic anemia; r
- normocytic normochromic anemia; ref
- nystagmus; Nystag
- peripheral neuropathy; PCD
- paracentral retinal dysfunction; ppv
- pyramidal tract signs; Pyram
- sensory deficits; Sen
- transient; V
- reduction of paramacular cone function; figure 2C

**DISCUSSION**

The neurologic phenotype of ATXPC with **SAMD9L** mutations is characterized by nystagmus and slow progressive balance impairment. These signs were found in almost all **SAMD9L** mutation carriers examined in this study (table). The
Figure 2  Neuroimaging and multifocal electretinography (mfERG)

(A and B) Neuroradiologic findings in affected members of family 1 (A) and family 2 (B). Age at examination is provided in parentheses. pHSCT, examination performed posthematopoeitic stem cell transplantation for hematologic malignancy. *Also carry the rare variant SAMD9L c.689C>A in trans. Sagittal MRIs reveal cerebellar atrophy in all individuals examined in adult age. Patient F1: V-1 had cerebellar atrophy at 5 years of age, (A) but patient F2: II-1 did not have clear cerebellar atrophy at 5 years of age (not shown). Later, at the age of 12 years and 9 months F2: II-1 and F2: II-4 at the age of 7 years and 7 months, both had mild cerebellar atrophy (B). Bilateral hyperintense signal changes were visible in the frontoparietal periventricular white matter on T2-FLAIR images to a variable degree in all patients, except the 2 adults with the rare variant SAMD9L c.689C>A (A and B). Frontoparietal white matter changes of F2: II-4 and F2: II-1 decreased during childhood (B). F1: V-1 showed the most prominent white matter changes after hematologic malignancy and 3 months post-HSCT (A), but the white matter changes in F2: II-4, who also had hematologic malignancy and HSCT, decreased (B). For F1: V-1, the follow-up examinations 9 and 21 months post-HSCT showed unchanged widespread white matter abnormalities (not shown). FLAIR images revealed white matter abnormalities in the peritrigonal area in the individuals examined at 5 years and younger (B) that might be a sign of incomplete myelination normal to this age (see appendix e-1). Since there were also peritrigonal white matter signal abnormalities in the adult patient F2: I-2 (B), they might indicate vulnerability of these areas in SAMD9L-related ATXPC. CT of patient F1: II-3 showed cerebellar atrophy and diffuse periventricular hypodensities of the cerebral white matter (A). (C) mfERG of 2 patients, compared to normal findings in an adult, shows reduced paracentral function in both patients, to a different degree. N, normal; j, significantly reduced function. Fundus photography and optical coherence tomography showed normal results (appendix e-1). ATXPC = ataxia-pancytopenia; FLAIR = fluid-attenuated inversion recovery; ms and MS, milliseconds.

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neuro-otological findings indicate midline cerebellar dysfunction. Balance impairment was clearly visible on examination, but it was mild or very mild in some patients with only difficulty with tandem walking or standing in a tandem position. Additional signs include mild pyramidal signs, strabismus, and decreased vibration sense. Neuroimaging revealed marked cerebellar atrophy in all 8 patients and periventricular white matter changes in 6 of 8 (figure 2). Attention deficit hyperactivity disorder or cognitive problems were noticed in 4 patients, but it remains uncertain if these are related to the disorder.

Ophthalmological symptoms or signs have not previously been described for ATXPC with SAMD9L mutations. Adult patients from both families reported reading and focusing difficulties. mfERG showed intact function in the most central area but marked paracentral (cone) dysfunction, which correlates with these visual symptoms. Similar ophthalmologic findings have been reported, for example, in patients with SCA7. We suggest that some of the “atactic” signs in the upper extremities noted in patients with SAMD9L mutations may in fact be caused by a decrease in visual control of hand movements, rather than cerebellar ataxia.

In our as well as all previously reported families, all carriers of pathogenic SAMD9L mutations developed neurologic signs. Expressivity was variable from individuals experiencing marked balance impairment to those who only noticed mild problems with balance or gaze fixation. Careful neurologic examination, however, revealed clearly abnormal signs in all carriers with complete penetrance. The hematologic disorders in members of both families have previously been described in detail. In these and the other families, there was pancytopenia with a risk of severe hemorrhages or infections on the one hand, and myelodysplastic syndrome on the other hand, but these hematologic abnormalities were not noticed in all mutation carriers (table).

One of SAMD9L’s roles is that of a tumor suppressor or inhibitor of uncontrolled cell division. There is strong evidence that the disease-causing mutations exert a gain-of-function mechanism, resulting in more pronounced inhibition of cell division, which in blood cell lineages causes pancytopenia and genetic pressure to eliminate the mutant copy of the SAMD9L gene. Also in ataxia-telangiectasia, cerebellar degeneration co-occurs with immunologic deficiencies, lymphopenia, and increased risk of lymphoid malignancies. While dysfunction of DNA repair caused by mutations in ATM underlies some of the features of ataxia-telangiectasia, the cause of cerebellar degeneration in ataxia-telangiectasia remains difficult to explain. Perhaps similar to SAMD9L, ATM also influences cell-cycle signaling and cellular homeostasis pathways and interacts with another tumor suppressor, p53, in the cell-cycle control system. Cerebellar Purkinje cells and retinal cells might be particularly vulnerable to the effect of SAMD9L mutations.

**AUTHOR CONTRIBUTIONS**

Sorina Gorceenco: drafting/revising the manuscript for content, including medical writing for content, study concept or design, analysis or interpretation of data, acquisition of data, and other: examination of 13 adult patients, chart review. Jonna Komulainen-Ebrahim: drawing/revising the manuscript for content, including medical writing for content, analysis or interpretation of data, and other: clinical data on 2 Finnish pediatric patients. Katrin Nordborg: drafting/revising the manuscript for content, including medical writing for content, analysis or interpretation of data, and other: clinical data on 1 Swedish pediatric patient and sibling. Maria Suo-Palosaari: drafting/revising the manuscript for content, including medical writing for content, analysis or interpretation of data, and other: clinical data on 3 Finnish patients and interpretation of neuroradiologic data. Sten Andrénsson: drafting/revising the manuscript for content, including medical writing for content, analysis or interpretation of data, and other: clinical data on 1 Swedish pediatric patient and sibling. Maria Nordberg: drafting/revising the manuscript for content, including medical writing for content, analysis or interpretation of data, and other: clinical data on 1 adult patient. Christer Nilsson: revising the manuscript for content and other: clinical data on Swedish index patient (1 patient). Ulfika Kjellström: drafting/revising the manuscript for content, including medical writing for content, analysis or interpretation of data, and other: clinical data on 1 Swedish pediatric patient. Elisa Rahlkalla: revising the manuscript for content, including medical writing for content, analysis or interpretation of data, and other: clinical data on 1 Swedish pediatric patient. Mikael Karlberg: drafting/revising the manuscript for content, including medical writing for content, analysis or interpretation of data, and other: clinical data on HSCT treatment of 1 Swedish pediatric patient.

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