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Spinocerebellar Ataxia Type 17

[*SCA17*]

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Summary

Disease characteristics. Spinocerebellar ataxia type 17 (SCA17) is characterized by ataxia, dementia, and involuntary movements, including chorea and dystonia. Psychiatric symptoms, pyramidal signs, and rigidity are common. The age of onset ranges from three to 55 years. Individuals with full-penetrance alleles develop neurologic and/or psychiatric symptoms by age 50 years. Ataxia and psychiatric abnormalities are frequently the initial findings, followed by involuntary movement, parkinsonism, dementia, and pyramidal signs. MRI shows variable atrophy of the cerebrum, brain stem, and cerebellum. The clinical features correlate with the length of the CAA/CAG repeat.

Diagnosis/testing. The diagnosis of SCA17 relies on molecular genetic testing to detect an abnormal CAA/CAG repeat expansion in *TBP*, the only gene known to be associated with

SCA17. Affected individuals usually have more than 42 repeats. Such testing detects 100% of affected individuals.

Management. *Treatment of manifestations:* psychotropic medications for psychiatric problems, antiepileptic drugs for seizures (AEDs); botulinum toxin injections for dystonia; and adaptation of the environment to accommodate dementia. *Prevention of secondary complications:* Side effects of psychotropic medications and AEDs may require total or intermittent discontinuation of the treatment or reduction in dose. *Surveillance:* annual or semiannual evaluation by a neurologist or more frequently if symptoms are progressing rapidly. *Agents/circumstances to avoid:* Sedative/hypnotic agents, such as ethanol or certain medications, may exacerbate incoordination.

Genetic counseling. SCA17 is inherited in an autosomal dominant manner. Offspring of affected individuals have a 50% risk of inheriting the expanded *TBP* allele. The age of onset, severity, specific symptoms, and progression of the disease are variable and cannot be predicted by family history or size of expansion. Prenatal diagnosis is possible for fetuses at 50% risk if the diagnosis has been confirmed in at least one relative; requests for prenatal testing of typically adult-onset diseases are uncommon.

Diagnosis

Clinical Diagnosis

Spinocerebellar ataxia type 17 (SCA17) is suspected in individuals with the following:

- Psychiatric symptoms or dementia
- Cerebellar ataxia or involuntary movement

Molecular Genetic Testing

GeneReviews designates a molecular genetic test as clinically available only if the test is listed in the GeneTests Laboratory Directory *by either a US CLIA-licensed laboratory or a non-US clinical laboratory. GeneTests does not verify laboratory-submitted information or warrant any aspect of a laboratory's licensure or performance. Clinicians must communicate directly with the laboratories to verify information.*—ED.

Gene. *TBP*, encoding the TATA-box-binding protein, is the only gene associated with SCA17. The expansion of a CAA/CAG repeat is the only mutation observed [Koide et al 1999, Nakamura et al 2001].

Allele sizes. The structure of the repeat sequence is $(CAG)_3$ $(CAA)_3$ $(CAG)_x$ CAA CAG CAA $(CAG)_y$ CAA CAG.

- **Normal alleles:** 25 to 42 CAG/CAA repeats
- **Mutable normal alleles:** Not reported to date
- **Reduced penetrance alleles:** 43 to 48 CAA/CAG repeats. An individual with an allele in this range may or may not develop symptoms. The significance of alleles of 43 and 44 repeats is particularly controversial because penetrance is estimated to be 50%, making genotype-phenotype correlations difficult.
- **Full penetrance alleles:** 49 or greater CAA/CAG repeats. The largest repeat size reported to date is 66 [Maltecca et al 2003].

CAA CAG CAA interrupation. The CAA CAG CAA interruption between $(CAG)_x$ and $(CAG)_y$ is present in all expanded alleles that are stably transmitted (i.e., the allele size is unchanged during meiosis).

The CAA CAG CAA interruption between $(CAG)_x$ and $(CAG)_y$ was absent in two families with allele size instability (i.e., change in allele size) during transmission [Zuhlke et al 2001, Maltecca et al 2003]. Thus, loss of this interruption may be a prerequisite of instability in SCA17 as in other diseases caused by repeat expansions [Maltecca et al 2003; Zuhlke, Spranger et al 2003; Zuhlke et al 2005].

Clinical testing

- **Targeted mutation analysis.** Mutation analysis by direct amplification of the SCA17 CAA/CAG repeat identifies 100% of individuals who have a disease-causing *TBP* mutation.

Table 1 summarizes molecular genetic testing for this disorder.

Table 1. Molecular Genetic Testing Used in Spinocerebellar Ataxia Type 17

Test Method	Mutations Detected	Mutation Detection Frequency [1]	Test Availability
Targeted mutation analysis	CAA/CAG repeat expansion of *TBP*	100%	Clinical Testing

1. Proportion of affected individuals with a mutation(s) as classified by gene/locus, phenotype, population group, genetic mechanism, and/or test method

Testing Strategy

To establish the diagnosis in a proband, molecular genetic testing must reveal an expanded CAA/CAG repeat in the *TBP* gene.

Predictive testing for at-risk asymptomatic adult family members requires prior confirmation of the diagnosis in an affected family member.

Prenatal diagnosis for at-risk pregnancies requires prior confirmation of the diagnosis in an affected family member.

Genetically Related (Allelic) Disorders

No other phenotypes are associated with mutations in the *TBP* gene.

Clinical Description

Natural History

The main symptoms of spinocerebellar ataxia type 17 (SCA17) are ataxia (95%), dementia (~90%), and involuntary movements (~70%), including chorea and dystonia (blepharospasm, torticollis, writer's cramp, foot dystonia) [Toyoshima et al 2004b]. Psychiatric symptoms, pyramidal signs, and rigidity are common.

Onset ranges from age three to 55 years (mean: 33 years). All individuals with full penetrance alleles develop neurologic and/or psychiatric symptoms by age 50 years [Koide et al 1999; Fujigasaki et al 2001; Nakamura et al 2001; Zuhlke et al 2001; Silveira et al 2002; Maltecca et al 2003; Stevanin et al 2003; Zuhlke, Gehlken et al 2003; Bauer et al 2004; Hagenah et al 2004; Oda et al 2004; Toyoshima, personal observation].

Although the disease course is variable, ataxia and psychiatric abnormalities are frequently the initial findings followed by involuntary movement, parkinsonism, dementia, and pyramidal signs.

MRI shows variable atrophy of the cerebrum, brain stem, and cerebellum (Figure 1). Most people present with cerebellar atrophy. The age of the individual and the length of CAA/CAG

repeat influence the degree of atrophy. For example, in older individuals, even those with a small full-penetrance allele, severe atrophy is present on MRI. High-intensity T2-weighted images and selective atrophy on caudate nucleus are not observed. There is some correlation of region of brain atrophy with clinical characteristics [Lasek et al 2006].

Neuropathology. The brain shows atrophy of the striatum (more apparent in the caudate nucleus) and cerebellum. Histologically, neuronal loss is observed in the striatum and Purkinje cell layer. Loss of cerebral cortical neurons is seen in some individuals.

Immunohistochemistry for the expanded polyglutamine (polyQ) tracts shows diffuse labeling of the neuronal nucleoplasm. Note: Intranuclear inclusions are a much less common finding than diffuse labeling. No labeling is detectable in the cytoplasm or in the neuropil. Glial cell involvement is occasionally seen.

In individuals who are homozygous for an expanded allele in the full-penetrance range, nuclear polyQ pathology involves other CNS regions including the cerebral cortex, thalamus, and brain stem [Toyoshima et al 2004a]. The abundant nuclear accumulation of polyQ in the cerebral cortices and subcortical nuclei (e.g., dorsomedian thalamic nucleus) are possibly associated with the prominent cognitive and behavioral decline in affected individuals.

Genotype-Phenotype Correlations

Heterozygotes

Clinical features. The length of the CAA/CAG repeat correlates with the clinical features based on data available from 52 individuals (50 from the literature and two unreported) (Table 2). As the information reported in the literature was incomplete, the percentage of each symptom may be underestimated [Koide et al 1999;Fujigasaki et al 2001;Nakamura et al 2001;Zuhlke et al 2001;Silveira et al 2002;Maltecca et al 2003;Rolfs et al 2003;Stevanin et al 2003;Zuhlke, Gehlken et al 2003,Bauer et al 2004,Hagenah et al 2004,Oda et al 2004]. Of note is the high proportion of individuals with psychiatric problems and chorea.

- **CAA/CAG repeat size from 43 to 50.** More than 75% of individuals have intellectual deterioration; in some individuals intellectual problems and involuntary movements are the only signs. Psychiatric problems or dementia, parkinsonism, and chorea, a clinical constellation resembling Huntington disease, are more frequently observed in individuals with CAA/CAG repeats in this range than in individuals with larger repeats [Stevanin et al 2003, Bauer et al 2004, Toyoshima et al 2004a].
- **CAA/CAG repeat size from 50-60.** All individuals have ataxia and 75% have reduced intellectual function. Pyramidal signs (e.g., increased deep tendon reflexes) and dystonia are more common than in those with smaller repeats.
- **CAA/CAG repeat size greater than 60.** Two individuals with repeats in this size range have been reported. The largest CAA/CAG repeat is 66 repeats, observed in a familial case [Maltecca et al 2003]. The child developed gait disturbance at age three years followed by spasticity, dementia, and psychiatric symptoms. The other child, who had a *de novo* CAG repeat expansion of 63 repeats, developed ataxia and intellectual deterioration at age six years followed later by spasticity [Koide et al 1999]. MRI showed severe atrophy in the cerebrum, cerebellum, and brain stem.

Table 2. Frequency of Clinical Features in SCA17 Correlated with Repeat Size

CAA/CAG repeat size	Ataxia	Dementia or Psychiatric Symptoms	Increased DTRs [1]	Dystonia	Parkinsonism	Chorea
43-49	93%	90%	45%	7%	42%	39%
≥50	100%	75%	55%	40%	25%	10%

1. DTR = deep tendon reflex

See Figure 2.

Homozygotes. Two homozygous individuals and one compound heterozygous individual have been reported [Zuhlke, Spranger et al 2003; Oda et al 2004; Toyoshima et al 2004a]. Two individuals who were homozygous for 47 and 48 CAG/CAA repeats had onset in the fourth decade, not unlike the onset age predicted for heterozygotes [Zuhlke, Spranger et al 2003; Toyoshima et al 2004a]. The individuals' symptoms were severe and rapidly progressive, and in one case differed from those of his parents, suggesting that the presence of two expanded alleles influences the severity and rate progression of symptoms.

Penetrance

The penetrance of alleles of 43 to 44 repeats is estimated to be approximately 50% and the penetrance of alleles of 45 to 48 repeats is estimated to be greater than 80% [Toyoshima et al 2004a].

- An individual with 43 CAA/CAG repeats developed ataxia with dementia at age 52 years [Silveira et al 2002].
- An individual with 45 CAA/CAG repeats developed symptoms at age 60 years, the latest onset observed to date [Oda et al 2004].
- Asymptomatic elderly individuals with 43 to 49 CAA/CAG repeats have also been reported [Nakamura et al 2001; Zuhlke, Gehlken et al 2003; Oda et al 2004; Zuhlke et al 2005].

Age of onset. The correlation between the size of the CAG/CAA repeat and the age of onset in SCA17 (Figure 3) is not as strong as that in other disorders (SCA1, SCA2, SCA3, SCA6, SCA7, Huntington disease, DRPLA, SBMA [Kennedy disease]) caused by expansion of a polyglutamine tract [Rolfs et al 2003,Toyoshima et al 2004a].

Anticipation

Instability of the CAG repeat in germline transmission is not clear in SCA17 [Fujigasaki et al 2001, Nakamura et al 2001, Shatunov et al 2004].

The phenomenon termed anticipation, an earlier age of onset and more severe disease manifestations in offspring, is infrequently documented in families with SCA17. In addition, because of low penetrance of the intermediate alleles (43 to 48 repeats), the age of onset, severity, specific symptoms, and progression of the disease are variable and cannot be predicted by family history or size of expansion.

Prevalence

Fewer than 100 families with SCA17 have been reported.

The prevalence of SCA17 in the Japanese population is estimated at 0.47:1,000,000. SCA17 accounts for approximately 0.3% of autosomal dominant SCA [Maruyama et al 2002].

The minimum prevalence of SCA17 in northeast England is 0.16:100,000 [Craig et al 2005].

In a study of the Yugoslav population, none of the 115 individuals with autosomal dominant cerebellar ataxia or simplex cases of adult-onset ataxia had SCA17 [Alendar et al 2004].

The prevalence of SCA17 may be underestimated because some individuals with SCA17 have a phenotype similar to that of Huntington disease.

Differential Diagnosis

For current information on availability of genetic testing for disorders included in this section, see GeneTests Laboratory Directory. —ED.

The differential diagnosis of spinocerebellar ataxia type 17 (SCA17) includes many of the other hereditary ataxias that are summarized in the Hereditary Ataxia Overview and described in detail for specific ataxias, including SCA1, SCA2, SCA3, and SCA7.

DRPLA is a progressive disorder of ataxia, choreoathetosis, and dementia or character changes in adults and ataxia, myoclonus, epilepsy, and progressive intellectual deterioration in children.

Huntington disease (HD) needs to be considered [Rolfs et al 2003]. HD is a progressive disorder of motor, cognitive, and psychiatric disturbances. The mean age of onset is 35 to 44 years and the median survival is 15 to 18 years after onset.

Bauer and colleagues (2004) reported nine individuals with *TBP* alleles larger than 45 CAA/CAG repeats among 1,712 individuals with Huntington disease-like (HDL), and observed that CAA/CAG repeat expansions in the *TBP* gene represented a more common monogenic cause for a HD-like phenotype than Huntington disease-like 1 (HDL-1) [Xiang et al 1998] or Huntington disease-like 2 (HDL-2) [Margolis et al 2001].

Lin and colleagues (2006) reported an individual with SCA17 whose findings of ataxia, autonomic dysfunction, parkinsonism, supranuclear palsy, and cognitive impairment resembled multiple system atrophy.

Management

Evaluations Following Initial Diagnosis

To establish the extent of disease in an individual diagnosed with spinocerebellar ataxia type 17 (SCA17), the following evaluations are recommended:

- Neuropsychological testing to evaluate for dementia and/or psychiatric disturbance
- Brain MRI to evaluate areas and amount of atrophy

Treatment of Manifestations

- Treatment of psychiatric problems with appropriate psychotropic medications
- Treatment of seizures with antiepileptic drugs (AEDs)
- Adaptation of environment and care to the level of dementia
- Treatment of dystonia with local injections of botulinum toxin

Prevention of Secondary Complications

The side effects of psychotropic medications and AEDs (e.g., depression, sedation, nausea, restlessness, headache, neutropenia, and tardive dsykinesia) can be major secondary complications in persons with SCA17. For some individuals, the side effects of certain

therapeutics may be worse than the symptoms of the disease; such individuals may benefit from total or intermittent discontinuation of the treatment or reduction in dose.

Surveillance

Affected individuals should be followed annually or semiannually by a neurologist or more frequently if symptoms are progressing rapidly, as may happen in the advanced stages [Toyoshima et al 2004a].

Agents/Circumstances to Avoid

Agents with sedative/hypnotic properties, such as ethanol or certain medications, may markedly increase incoordination.

Testing of Relatives at Risk

See Genetic Counseling for issues related to testing of at-risk relatives for genetic counseling purposes.

Therapies Under Investigation

Search ClinicalTrials.gov for access to information on clinical studies for a wide range of diseases and conditions. Note: There may not be clinical trials for this disorder.

Other

Genetics clinics, staffed by genetics professionals, provide information for individuals and families regarding the natural history, treatment, mode of inheritance, and genetic risks to other family members as well as information about available consumer-oriented resources. See the GeneTests Clinic Directory.

Support groups have been established for individuals and families to provide information, support, and contact with other affected individuals. The Resources section may include disease-specific and/or umbrella support organizations.

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Genetic Counseling

Genetic counseling is the process of providing individuals and families with information on the nature, inheritance, and implications of genetic disorders to help them make informed medical and personal decisions. The following section deals with genetic risk assessment and the use of family history and genetic testing to clarify genetic status for family members. This section is not meant to address all personal, cultural, or ethical issues that individuals may face or to substitute for consultation with a genetics professional. To find a genetics or prenatal diagnosis clinic, see the GeneTests Clinic Directory.

Mode of Inheritance

Spinocerebellar ataxia type 17 (SCA17) is inherited in an autosomal dominant manner.

Risk to Family Members

Parents of a proband *

- Approximately 50% of individuals diagnosed with SCA17 have an affected parent.
- Thirty-eight percent of individuals with SCA17 are simplex cases (i.e., only one affected person in the family). In most cases, molecular genetic testing of the parents has not been performed.
- A proband with SCA17 may have the disorder as a result of a *de novo* expansion in the *TBP* gene. The proportion of cases caused by *de novo* expansions is unknown [Koide et al 1999, Shatunov et al 2004].
- If an expansion (>42 CAA/CAG repeats) of the *TBP* gene cannot be detected in the DNA of either parent, two possible explanations are germline mosaicism in a parent or a *de novo* expansion in the proband. Expansion from a mutable normal allele in the parent is also a possibility, although no instances have been reported.
- Recommendations for the evaluation of parents of a proband with an apparent *de novo* mutation include molecular genetic testing of the *TBP* gene.

Note: Although approximately 50% of individuals diagnosed with SCA17 have an affected parent, the family history may appear to be negative because of failure to recognize the disorder in family members because of its extremely variable phenotype, early death of the parent before the onset of symptoms, late onset of the disease in the affected parent, or reduced penetrance in a parent.

* Based on the family history of the 37 reported families with affected individuals and an unreported family with two affected individuals [Koide et al 1999; Fujigasaki et al 2001; Nakamura et al 2001; Zuhlke et al 2001; Silveira et al 2002; Maltecca et al 2003; Rolfs et al 2003; Stevanin et al 2003; Zuhlke, Gehlken et al 2003; Bauer et al 2004; Hagenah et al 2004; Oda et al 2004; Toyoshima, personal observations]

Sibs of a proband

- The risk to the sibs of the proband depends upon the genetic status of the proband's parents.
- If one of the parents of the proband has an expanded *TBP* allele, the risk to the sibs of inheriting the expanded CAA/CAG allele is 50%. The age of onset, severity, specific symptoms, and progression of the disease are variable and cannot be precisely predicted by family history or size of expansion.
- If an expansion (>42 CAA/CAG repeats) of the *TBP* gene cannot be detected in the DNA of either parent, the risk to sibs is low but greater than that of the general population because of the possibility of germline mosaicism.

Offspring of a proband

- Each child of an individual with SCA17 has a 50% chance of inheriting the expanded *TBP* allele.
- The age of onset, severity, specific symptoms, and progression of the disease are variable and cannot be precisely predicted by family history or size of expansion.

Other family members of a proband. The risk to other family members depends upon the genetic status of the proband's parents. If a parent is found to be affected or to have an expanded *TBP* allele, his or her family members are at risk.

Related Genetic Counseling Issues

Considerations in families with an apparent *de novo* mutation. When neither parent of a proband with an autosomal dominant condition has gene expansion or clinical evidence of the disorder, it is likely that the proband has a *de novo* mutation. However, possible non-medical explanations including alternate paternity or undisclosed adoption could also be explored.

Family planning. The optimal time for determination of genetic risk and discussion of the availability of prenatal testing is before pregnancy. Similarly, decisions about testing to determine the genetic status of at-risk asymptomatic family members are best made before pregnancy.

Testing of at-risk asymptomatic adults. Testing of asymptomatic adults at risk for SCA17 is available using the same techniques described in Molecular Genetic Testing. Such testing is not useful in predicting age of onset, severity, type of symptoms, or rate of progression in asymptomatic individuals. When testing at-risk individuals for SCA17, an affected family member should be tested first to confirm the molecular diagnosis of SCA17 in the family.

Testing for the disease-causing expansion in the absence of definite symptoms of the disease is predictive testing. At-risk asymptomatic adult family members may seek testing in order to make personal decisions regarding reproduction, financial matters, and career planning. Others may have different motivations including simply "the need to know."

Testing of asymptomatic at-risk adult family members usually involves pretest interviews in which the motives for requesting the test, the individual's knowledge of SCA17, the possible impact of positive and negative test results, and neurologic status are assessed. Those seeking testing should be counseled about possible problems that they may encounter with regard to health, life, and disability insurance coverage, employment and educational discrimination, and changes in social and family interaction. Another issue to consider is the implications for the at-risk status of other family members. Informed consent should be procured and records kept confidential. Individuals with a positive test result need arrangements for long-term follow-up and genetic counseling.

Testing of at-risk asymptomatic individuals during childhood. Consensus holds that individuals younger than age 18 years at risk for adult-onset disorders should not have testing in the absence of symptoms. The principal arguments against such testing are that it removes the individual's choice to know or not know this information, it raises the possibility of stigmatization within the family and in other social settings, and it could have serious educational and career implications. Individuals younger than 18 years of age who are symptomatic usually benefit from having a specific diagnosis established. (See also the National Society of Genetic Counselors points to consider: ethical, legal, and psychosocial implications of genetic testing in children and adolescents (see Genetic Testing).

DNA banking. DNA banking is the storage of DNA (typically extracted from white blood cells) for possible future use. Because it is likely that testing methodology and our understanding of genes, mutations, and diseases will improve in the future, consideration should be given to banking DNA of affected individuals. See DNA Banking for a list of laboratories offering this service.

Prenatal Testing

Prenatal diagnosis for pregnancies at increased risk is possible by analysis of DNA extracted from fetal cells obtained by amniocentesis usually performed at about 15-18 weeks' gestation or chorionic villus sampling (CVS) at about ten to 12 weeks' gestation. The disease-causing allele of an affected family member must be identified before prenatal testing can be performed.

Note: Gestational age is expressed as menstrual weeks calculated either from the first day of the last normal menstrual period or by ultrasound measurements.

Requests for prenatal testing for (typically) adult-onset conditions such as SCA17 are not common. Differences in perspective may exist among medical professionals and within families regarding the use of prenatal testing. Although most centers would consider decisions about prenatal testing to be the choice of the parents, careful discussion of these issues is appropriate.

Preimplantation genetic diagnosis (PGD) may be available for families in which the disease-causing mutation has been identified in an affected family member. For laboratories offering PGD, see Testing.

Molecular Genetics

Information in the Molecular Genetics tables is current as of initial posting or most recent update. —ED.

Table A. Molecular Genetics of Spinocerebellar Ataxia Type17

Gene Symbol	Chromosomal Locus	Protein Name
TBP	6q27	TATA-box-binding protein

Data are compiled from the following standard references: Gene symbol from HUGO; chromosomal locus, locus name, critical region, complementation group from OMIM; protein name from Swiss-Prot.

Table B. OMIM Entries for Spinocerebellar Ataxia Type17

600075	TATA BOX-BINDING PROTEIN; TBP
607136	SPINOCEREBELLAR ATAXIA 17; SCA17

Table C. Genomic Databases for Spinocerebellar Ataxia Type17

Gene Symbol	Entrez Gene	HGMD
TBP	6908 (MIM No. 600075)	TBP

For a description of the genomic databases listed, click here.

Normal allelic variants: *TBP* consists of eight exons. The CAG/CAA repeat in the *TBP* gene is located in exon 3 and is predicted to code for a polyglutamine stretch. The CAG/CAA repeat size in normal individuals ranges from 25 to 42 repeats.

Pathologic allelic variants: The CAG/CAA repeat size in individuals with SCA17 ranges from 43 to 66 repeats.

Normal gene product: TATA-box-binding protein (TBP) is also called transcription factor IID (TFIID). Human TBP has 339 amino acids and a molecular size of 37.8 kd. TBP is an important general transcription initiation factor and is the DNA-binding subunit of RNA polymerase II transcription factor D, the multi-subunit complex crucial for the expression of most genes.

Abnormal gene product: Because TBP is a fundamental transcription factor expressed ubiquitously in all organs including the CNS, the question of whether loss of TBP function plays a role in the pathogenesis of SCA17 remains to be addressed. In a homozygote, however, no abnormality had been observed in growth, and pathologic examination had shown no specific changes in the visceral organs [Toyoshima et al 2004a]. Taking into consideration the

ubiquitous presence of TBP, the selective neuronal degeneration suggests no significant loss of protein function in individuals with SCA17.

Resources

GeneReviews provides information about selected national organizations and resources for the benefit of the reader. GeneReviews is not responsible for information provided by other organizations. Information that appears in the Resources section of a GeneReview is current as of initial posting or most recent update of the GeneReview. Search GeneTests*for this disorder and select* Resources *for the most up-to-date Resources information.*—ED.

NCBI Genes and Disease
Spinocerebellar ataxia

Spinocerebellar Ataxia: Making an Informed Choice about Genetic Testing
Booklet providing information about spinocerebellar ataxia
depts.washington.edu/neurogen/SpinoAtaxia.pdf

euro-ataxia (European Federation of Hereditary Ataxias)
Boherboy Dunlavin
Co Wicklow
Ireland
Phone: 045 401218
Fax: 045 401371
Email: mary.kearneyl@euro-ataxia.org
www.euro-ataxia.org

International Network of Ataxia Friends (INTERNAF)
www.internaf.org

National Ataxia Foundation
2600 Fernbrook Lane Suite 119
Minneapolis MN 55447
Phone: 763-553-0020
Fax: 763-553-0167
Email: naf@ataxia.org
www.ataxia.org

WE MOVE (Worldwide Education and Awareness for Movement Disorders)
204 West 84th Street
New York NY 10024
Phone: 800-437-MOV2 (800-437-6683)
Fax: 212-875-8389
Email: wemove@wemove.org
www.wemove.org

References

Medical Genetic Searches: A specialized PubMed search designed for clinicians that is located on the PubMed Clinical Queries page. PubMed

Published Statements and Policies Regarding Genetic Testing

No specific guidelines regarding genetic testing for this disorder have been developed.

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Chapter Notes

Revision History

- 1 August 2007 (me) Comprehensive update posted to live Web site
- 29 March 2005 (me) Review posted to live Web site
- 24 August 2004 (yt) Original submission

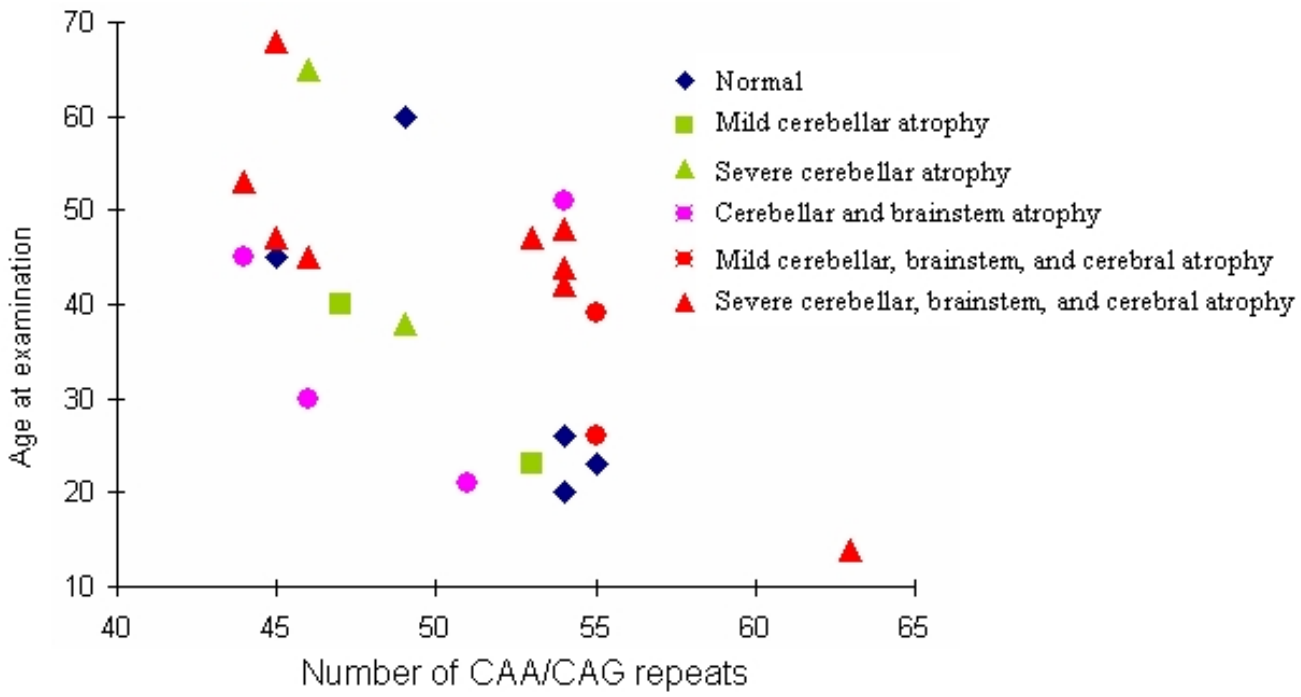

Figure 1. Number of CAA/CAG repeats versus age of individuals with SCA17

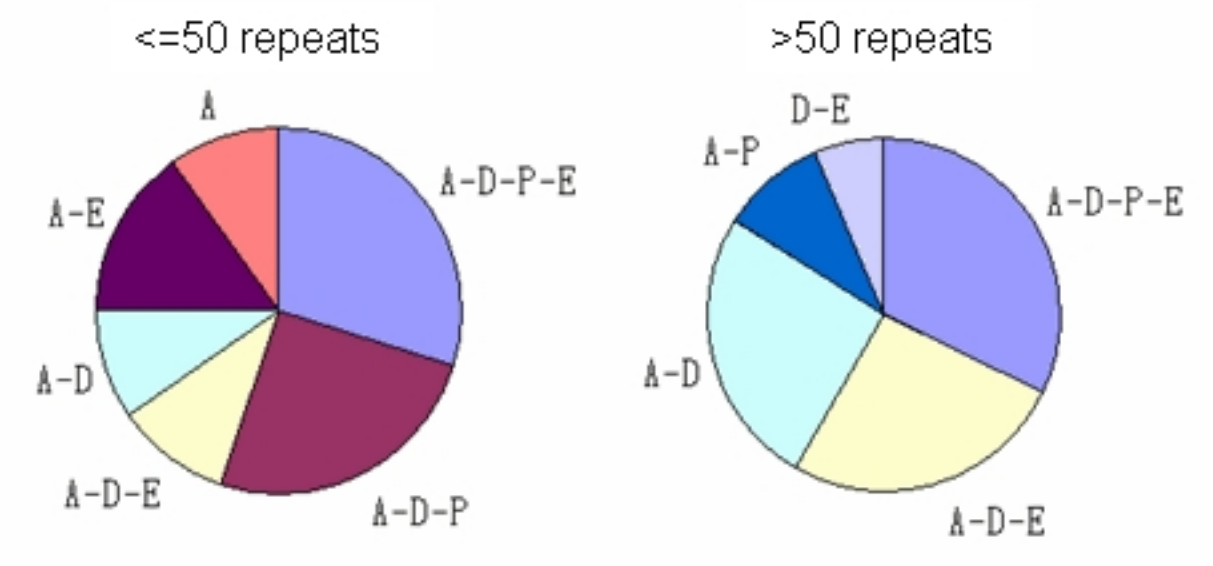

Figure 2. The clinical features in SCA17 depend on the length of CAA/CAG repeats. The clinical features of SCA17 in each case are denoted by letter. For example, A-E denotes ataxia with parkinsonism.
A = ataxia
D = dementia or psychiatric symptoms
P = pyramidal signs
E = parkinsonism or involuntary movement

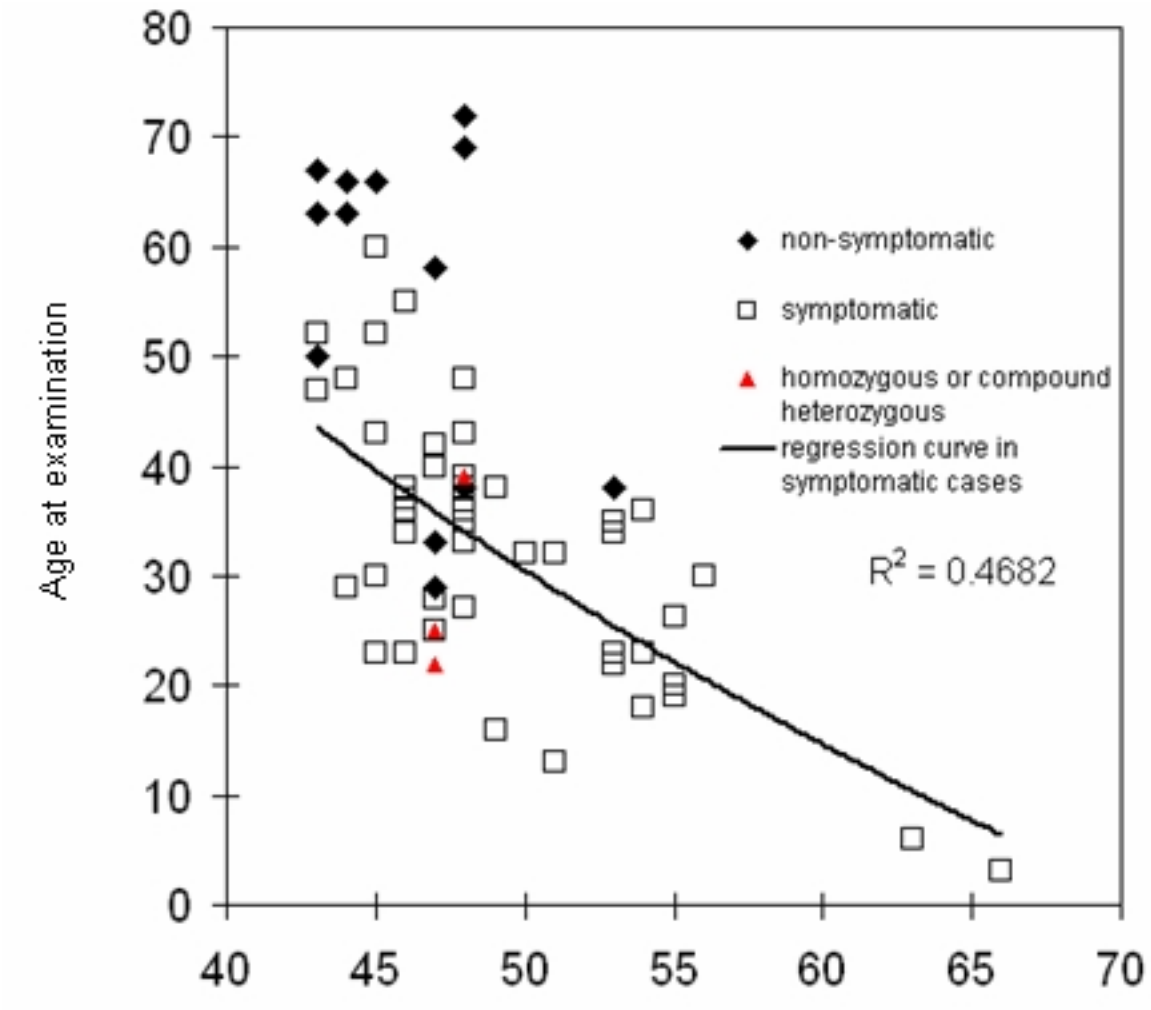

Figure 3. Correlation in SCA17 between age at onset and length of CAA/CAG repeat