Specific Treatments for Genetic Ataxias

INFORMATION FROM PATIENTS AND FAMILIES
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Some **genetic ataxias** have specific treatments. These include the following:

1. **Friedreich's ataxia (FA):** Omaveloxolone is a drug for FA and has been approved for patients aged 16 years or older in some countries. It has been shown to slow the progression of physical impairment (coordination, walking, balance) in FA though it is not a curative treatment. It is not yet available n India except on research basis.
2. **Ataxia with vitamin E deficiency (AVED):** AVED is a genetic ataxia caused by a mutation in the TTPA gene, resulting in very low levels of vitamin E in nervous tissues. It is treated with high-dose oral supplementation with vitamin E.
3. **Abetalipoproteinemia:** Abetalipoproteinemia is rare metabolic disorder that causes ataxia and recurrent diarrhoea. It is treated with low-fat diet and vitamin supplementation (e.g., vitamin E, A).
4. **Primary biotinidase deficiency:** Biotinidase deficiency is an inherited metabolic disorder caused by mutations in the BTD gene. The enzyme biotinidase normally helps recycle the vitamin biotin so that it can be used by various carboxylase enzymes in the body. Oral biotin supplementation is used for the treatment of this ataxia.
5. **Cerebrotendinous xanthomatosis (CTX):** CTX is a rare genetic disorder in which ataxia may occur. Chenodeoxycholic acid (a bile acid replacement) is used for the treatment of CTX. It helps to reduce the accumulation of harmful metabolites. However, this drug is currently unavailable in India.
6. **Spinocerebellar ataxia type 27B and 27A:** SCA27B is a late-onset genetic ataxia caused by intronic repeat expansion in the FGF14 gene. Similarly, the FGF14 gene mutation can also lead to SCA27A, which can present with early-onset tremor with late-onset progressive ataxia. Both may show a fluctuating course. 4-aminopyridine or acetazolamide may help to reduce the severity of episodes of ataxia.
7. **Episodic ataxia type 2:** EA2 is caused by genetic mutation in the CACNA1A gene, in which acetazolamide and 4- aminopyridine help to reduce the severity and frequency of episodes of ataxia.
8. **Niemann-Pick's disease type C (NPC):** NPC is a genetic lipid storage disorder which can present with ataxia. In some countries, the drug miglustat has been used to slow the progression of disease. Miglustat is currently unavailable in India.
9. **Coenzyme Q10 deficiency:** Some inherited ataxias due to CoQ10 (ubiquinone) deficiency are treated with high-dose oral CoQ10 supplementation.
10. **Glucose Transporter Type 1 Deficiency Syndrome (GLUT1 deficiency):** In GLUT1 deficiency, although classically seizures are seen, paroxysmal movement disorders and ataxia may also occur. Dietary ketogenic therapy may reduce symptoms in patients with GLUT1 deficiency disorders.
11. **Maple Syrup Urine Disease (MSUD):** MSUD is a genetic disorder that can present with intermittent ataxia. Low protein and leucine-restricted diet is helpful in this disorder.
12. **Refsum disease:** Refsum disease can present with ataxia, skin changes and hearing loss. A diet reduced in phytanic acid is helpful.
13. **Cerebral Folate Deficiency:** Cerebral Folate Deficiency is caused by a mutation in the FOLR1 gene and causes ataxia and seizures. Folinic acid supplementation is the treatment of choice.