**Centre for Arab Genomic Studies**
**A Division of Sheikh Hamdan Award for Medical Sciences**

The Catalogue for Transmission Genetics in Arabs
**CTGA Database**

# Spastic Paraplegia 24, Autosomal Recessive

## Alternative Names
SPG24

## Record Category
Disease phenotype

## WHO-ICD
Diseases of the nervous system > Systemic atrophies primarily affecting the central nervous system

## Incidence per 100,000 Live Births
Unknown

## OMIM Number
607584

## Mode of Inheritance
Autosomal recessive

## Gene Map Locus
13q14

## Description
The Hereditary spastic paraplegias form a group of genetically and clinically heterogeneous disorders characterized by progressive weakness and spasticity of the lower extremities. SPG24 is a pure or uncomplicated form of SPG, as no other neurological abnormalities are associated with the disorder. Symptoms usually manifest around 1-year of age and include difficulty walking, clonus, scissoring spastic gait, hyperreflexia and walking on tiptoes. It is found to affect both men and women.

Diagnosis is based on clinical presentation. Currently there is no treatment for SPG. Management of the disorder includes physical therapy, the use of orthotic braces to help foot drop and drugs such as baclofen to help relieve spasticity. SPG does not affect life expectancy and patients can live independent lives with the help of walking aids or wheelchairs.

## Molecular Genetics
Unlike most forms of the disorder, SPG24 has been shown to follow an autosomal recessive pattern of inheritance. Homozygosity mapping and linkage analysis have mapped the syndrome to chromosome 13q14 spanning a region of 1.8 Mb. However, the exact genetic defect leading to the syndrome has not yet been uncovered.

## Epidemiology in the Arab World

### Saudi Arabia

Hodgkinson et al. (2002) carried out a genetic analysis on a northern Saudi Arabian, consanguineous family with autosomal recessive spastic paraplegia affected members. The healthy parents had 9 children: 2 with spastic paraplegia, 2 with prelingual sensorineural deafness and 3 with both spastic paraplegia and deafness. Paraplegia affected patients showed symptoms around 1-year of age. Symptoms progressed to plantar flexion and inversion of both feet with sustained clonus, spastic scissoring gait, mild toe walking and crushing position at the knee. None of the family members showed any mental deficits and metabolic, cardiac and biochemical investigations were all normal. Whole genome scan, heteroduplex analysis and linkage analysis were able to identify a 1.8 Mb region of homozygosity on chromosome 13 in SPG affected individuals. The locus was flanked by the FLJ11712 gene and the D13S270 marker and had a LOD score of 3.26. The deafness trait was not found to co-segregate with the trait of paraplegia confirming that it is a separate condition independent of SPG.

## References
Hodgkinson CA, Bohlega S, Abu-Amero SN, Cupler E, Kambouris M, Meyer BF, Bharucha VA. A novel form of autosomal recessive pure hereditary spastic paraplegia maps to chromosome 13q14. Neurology. 2002; 59(12):1905-9.

## Related CTGA Records

## External Links
http://rarediseases.org/rare-diseases/hereditary-spastic-paraplegia/
http://emedicine.medscape.com/article/306713-overview

## Contributors
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