

[Cell Lines](#) [SACS/ARSACS/SPAX6](#) [Publication](#)

BOR-090711-003

IDENTIFICATION

| | |
|-------------------------|---|
| Causal gene(s) | SACS |
| Repeat size or mutation | Protein change: p.L1180Lfs*7 (nonsense compound heterozygous variant) |
| Cell type | Fibroblast |

DONOR INFORMATION

| | |
|------------------------------|----|
| Age at disease onset (years) | 25 |
| Age at collection (years) | 48 |

SOURCE & PUBLICATIONS

| | |
|---|--|
| Originating lab / institution | University of Bordeaux |
| Links to publications or public resources | New practical definitions for the diagnosis of autosomal recessive spastic ataxia of Charlevoix-Saguenay - PubMed https://pubmed.ncbi.nlm.nih.gov/26288984/ |