Cell Lines SACS/ARSACS/SPAX6 Publication

# AFT-068-005

## IDENTIFICATION

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|---|---|
| **Causal gene(s)** | SACS |
| **Repeat size or mutation** | Protein change: p.F2056Ffs*21 (nonsense+missense heterozygous variant) |
| **Cell type** | Fibroblast |

## DONOR INFORMATION

| | |
|---|---|
| **Age at disease onset (years)** | 1 |
| **Age at collection (years)** | 53 |

## SOURCE & PUBLICATIONS

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|---|---|
| **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<br>https://pubmed.ncbi.nlm.nih.gov/26288984/ |