# DISEASE:
# Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome

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| **NAME:** | Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome |
| **DESCRIPTION:** | Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome is a rare, genetic, syndromic intellectual disability disorder characterized by severe intellectual disability, non-inherited, progressive, post-natal microcephaly, hypotonia, hyperkinesia, absence of speech, strabismus, and midline stereotypic hand movements (e.g. hand washing/rubbing). Additional features include developmental delay, seizures and behavioral disturbances, such as self injury and unexplained crying episodes. |
| **ORPHACODE:** | 397933 |
| **SYNONYMS:** | IQSEC2-related syndromic intellectual disability |
| **XREF(S):** | Orphanet<br>ICD-10 |
| **ANALYTE(S):** | IQSEC2 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

Source URL: http://gentest.healthdata.be/disease/2364

# RELATED CONTENT

## Related Genetic Tests

- Epilepsy (gene panel)

## Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

## Related Analytes

- IQ motif and Sec7 domain ArfGEF 2

## Related Gene Panels

- Rare epilepsy with developmental delay (> 240 genes) - UZA