# DISEASE:
# Ichthyosis-short stature-brachydactyly-microspherophakia syndrome

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| **NAME:** | Ichthyosis-short stature-brachydactyly-microspherophakia syndrome |
| **DESCRIPTION:** | A rare, syndromic ichthyosis characterized by a collodion membrane at birth, generalized congenital ichthyosis, microspherophakia, myopia, ectopia lentis, short stature with brachydactyly and joint stiffness, and occasionally mitral valve dysplasia. |
| **ORPHACODE:** | 363992 |
| **SYNONYMS:** | 15q26.3 microdeletion syndrome |
| **XREF(S):** | Orphanet<br>ICD-10<br>OMIM |
| **ANALYTE(S):** | ADAMTS17<br>CERS3 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Ichthyosis (gene panel)

## Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- ADAM metallopeptidase with thrombospondin type 1 motif 17
- ceramide synthase 3

## Related Gene Panels

- Ichthyosis and erythroderma (98 genes) - KUL

Source URL: http://gentest.healthdata.be/disease/1836