# DISEASE:
# Autosomal dominant aplasia and myelodysplasia

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| **NAME:** | Autosomal dominant aplasia and myelodysplasia |
| **DESCRIPTION:** | A rare, genetic, hematologic disorder characterized by bone marrow failure which manifests with aplastic anemia and/or myelodysplasia, associated with hearing/ear abnormalities (such as deafness, labyrinthitis), inherited in an autosomal dominant manner. |
| **ORPHACODE:** | 314399 |
| **SYNONYMS:** | Autosomal dominant aplastic anemia and myelodysplasia |
| **XREF(S):** | Orphanet<br>ICD-10<br>OMIM |
| **ANALYTE(S):** | SRP72 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Myeloid neoplasms with germline predisposition (Hereditary MDS/Acute Leukemia) (gene panel)
- « Inherited bone marrow failures syndromes » with or without organ dysfunction

## Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman

## Related Analytes

- signal recognition particle 72

## Related Gene Panels

- Hematologic Familiar Forms - ULG
- Hereditary Myelodysplastic /Acute Leukemia Predisposition Syndromes (gene panel)