# DISEASE:
# Dowling-Degos disease

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| **NAME:** | Dowling-Degos disease |
| **DESCRIPTION:** | A rare, genetic, hyperpigmentation of the skin disease characterized by adulthood-onset of reticular, reddish-brown to dark-brown, macular and/or comedone-like, hyperkeratotic papules with hypopigmented macules, predominantly affecting flexural areas and, on occasion, progressing to involve trunk and acral regions. Histologically, epidermal acanthosis, thin, branch-like, rete ridges, and a tendency for acantholysis and pigmentary incontinence is observed. |
| **ORPHACODE:** | 79145 |
| **SYNONYMS:** | Reticular pigment anomaly of flexures |
| **XREF(S):** | Orphanet<br>OMIM<br>OMIM<br>OMIM<br>OMIM<br>MedDRA<br>ICD-10 |

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| **ANALYTE(S):** | PSENEN<br>KRT5<br>POFUT1<br>POGLUT1 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

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# RELATED CONTENT

## Related Genetic Tests

- Ichthyosis (gene panel)
- Periodic Fever (88 genes)

## Related Laboratories

- Centre de Génétique Humaine - Erasme ULB
- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- keratin 5
- protein O-fucosyltransferase 1
- protein O-glucosyltransferase 1
- presenilin enhancer, gamma-secretase subunit

## Related Gene Panels

- Ichthyosis and erythroderma (98 genes) - KUL
- Periodic Fever (88 genes) - ULB