# Leprechaunism Syndrome

**Eponyms:**
- Donohue syndrome
- insulin receptor defect
- insulin resistance severe

**Inheritance:**
- autosomal recessive

**Semiological Synthesis:**
Cutaneous-facio-endocrino-genital disorder. Severe dystrophy, low birth weight, elfin face, mental retardation, failure to thrive, prominent clitoris/penis/ears/hands and feet, hypoglycemia, severe insulin resistance, visceral anomalies; occasionally Horner syndrome.

<table>
<thead>
<tr>
<th>Group</th>
<th>Sub group</th>
<th>Signs:</th>
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<tbody>
<tr>
<td>DERMATOLOGICAL DISORDERS</td>
<td></td>
<td>cutis, changes in appearance and/or features</td>
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<td></td>
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<td>cutis, hyperelastic</td>
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<td></td>
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<td>cutis, redundant, loose, including cutis laxa</td>
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<td>hand, redundant cutis</td>
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<td></td>
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<td>cutis, nodules</td>
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<td>lipodystrophy</td>
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<td>keratinisation defects</td>
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<td></td>
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<td>acanthosis nigricans</td>
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<td></td>
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<td>lanugo, modified appearance</td>
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<td></td>
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<td>hypertrichosis, hirsutism</td>
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<td>subcutaneous changes</td>
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<td>subcutaneous, scarce, thin</td>
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| ENDOCRINOLOGICAL, METABOLIC DISORDERS | | pancreas, islet modified functions |
| | | pancreas, islet dysfunction |

| GENITAL DISORDERS | | breast, changes |
| | | breast, early development |
| | | female genitalia, modified not including ambiguity |
| | | clitoris, hypertrophic, clitoromegaly |
| | | genital dysfunctions |
| | | hypogonitilism, hypogonadism; small testes, microorchidism, hypoplastic scrotum |
| | | male genitalia, modifications not including ambiguity |
| | | cryptorchidism |
| | | macropenis |

| LABORATORY DATA | | biochemical markers |
| | | metabolic defect |
| | | chromosomal assignment |
| | | chromosome 19p localization |
| | | gene, structural-functional abnormalities |
| | | gene analysis-DNA analysis |
| | | INSR insulin receptor, gene 19p13.2 |
| | | pancreatic hormones, modified functions |
| | | insulin, high levels; hyperinsulinemia |
| | | insulin-resistance |
| | | plasma nonprotein-organic constituents, anomalies |
| | | aminoacidemia, unspecified type |
| | | carbohydrates disorders |
| | | diabetes mellitus in syndromic association |
| | | hyperglycemia |
| | | hypoglycemia |
| | | tissue, biochemical changes |
| | | metabolic acidosis, ketonuria, ketosis, ketonemia, ketoacidosis |

**MUSCULAR DISEASES**
muscular defects, distress rectal
- diastasis recti
- hernia, inguinal
- hernia, umbilical, navel
systemic muscular defects
- hypotonia, muscular weakness, muscular hypoplasia, myastenia, not including atrophy
- limpness, severe hypotonia

NEUROLOGICAL DISORDERS
brain anomalies
- microcephaly, microcrania
mental retardation
- mental retardation

OROCRANIOFACIAL ANOMALIES
external ear malformations
- ear lobe, large, flare
- ear, low set
- ear, prominent; ear lobe, protruding
facies, modified appearance
- dysmorphic face
- facial dysmorphism due to cranial changes including microcephaly
- facies, coarse
- facies, pointed facies, elfin-like
- facies, senile, folded progeroid facies, premature aging, pinched
- proptosis, prominent eyes, exophthalmos, protuberant eyes, protruding eyes
forehead-orbital region, changes
- forehead, hairy, hypertrichosis
- frontal bossing
- hypertelorism
lips, modified appearance
- lips, protruding, everted, full, thickened
- lower lip, thick
mouth, modified appearance
- mouth, large, wide mouth, macrostomia
nose, modified appearance
- nose, large, broad, bulbous
oral mucous membranes, changes
- gingival hyperplasia, gum hypertrophy, broad alveolar ridge
teeth, modified structures
- teeth, carious
longue, changes
- macroglossia

OTHERS
inheritance
- inheritance, autosomal recessive
supergroups
- cutaneous-genito-neuro-skeletal disorders
- endocrino-neuro disorders
- facio-genito-neuro-skeletal disorders

PRENATAL-NEONATAL MODIFIED DATA
foetal changes
- birth length, decreased; low birth length
- birth weight, low; foetal growth decreased, intrauterine growth retardation, intrauterine growth restriction, IUGR
- floppy baby
- foetal changes, recognized by laboratory data
- foetal changes, recognized by ultrasound techniques

leprechaunism syndrome
prenatal diagnosis
- prenatal diagnosis, echographic
- prenatal diagnosis, metabolic
- prenatal diagnosis, molecular

SKELETAL DISORDERS
hand-foot, changes
- hand, large, broad
limb anomalies, limb defects
- lethality, in malformations, no
chondrodystrophy, no chromosomopathies
ossification, changes
- bone age, delayed
 stature, growth, modified habitus
- growth delayed, failure to thrive, growth
retardation

Super group: complex plurimutations
- cutaneous-genito-neuro-skeletal disorders
dysmorphic face-mental retardation
- endocrin-neuro disorders
- facio-genito-neuro-skeletal disorders

Super aggrev. Aggregations: FOETAL CHANGES
- foetal changes, recognized by laboratory data
- foetal changes, recognized by ultrasound techniques

LETHALITY
- lethality in plurimutations, not including chondrodystrophy or chromosomopathies

METABOLIC DISORDERS
- carbohydrates, disorders
- metabolic acidosis, ketonuria, ketosis, ketonemia, ketoacidosis

OTHER
- craniofacial dysmorphism due to cranial changes, including microcephaly
- diabetes, mellitus in syndromic association
- dysmorphic face
- floppy baby

Differential diagnosis:
85 acanthosis nigricans, Flier type
90 acanthosis nigricans, malignant type
80 acanthosis nigricans-diabetes
3140 Berardinelli-Seip syndrome
5930 Cockayne I syndrome
5931 Cockayne II syndrome
7605 diencephalic syndrome
27200 foetal benzodiazepine effect
27973 foetal maternal Sjogren-Mikulicz syndrome
11945 hemochromatosis, neonatal
21590 progeria
21810 pseudoleprechaunism

Aggregation(s) [in differential diagnosis]:
- lethality, in chromosomal disorders

Bibliography
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