### 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.

#### Signs:

<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>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>hypertrichosis, hirsutism</td>
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<td>subcutaneous changes</td>
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<td>subcutaneous, scarce, thin</td>
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<td>ENDOCRINOLOGICAL, METABOLIC DISORDERS</td>
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<td>pancreas, islet modified functions</td>
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<td>pancreas, islet dysfunction</td>
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<td>GENITAL DISORDERS</td>
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<td>breast, changes</td>
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<td>breast, early development</td>
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<td>female genitalia, modified not including ambiguity</td>
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<td>clitoris, hypertrophic, clitoromegaly</td>
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<td>genital dysfunctions</td>
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<td>hypogonitalism, hypogonadism; small testes, microorchidism, hypoplastic scrotum</td>
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<td>male genitalia, modifications not including ambiguity</td>
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<td>cryptorchidism</td>
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<td>macrogenus</td>
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<td>LABORATORY DATA</td>
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<td>biochemical markers</td>
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<td>metabolic defect</td>
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<td>chromosome 19p localization</td>
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<td>gene, structural-functional anomalies</td>
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<td>INSR insulin receptor, gene 19p13.2</td>
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<td>pancreatic hormones, modified functions</td>
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<td>insulin, high levels; hyperinsulinemia</td>
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<td>insulin-resistance</td>
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<td>plasma nonprotein-organic constituents, anomalies</td>
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<td>aminoacidemia, unspecified type</td>
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<td>carbohydrates disorders</td>
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<td>diabetes mellitus in syndromic association</td>
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<td>hyperglycemia</td>
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<td>hypoglycemia</td>
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<td>tissue, biochemical changes</td>
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<td>metabolic acidosis, ketonuria, ketosis, ketonemia, ketoacidosis</td>
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</tbody>
</table>

#### Muscular Diseases
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.
muscular defects, distrectual
  - 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 membrane, changes
  - gingival hyperplasia, gum hypertrophy, broad alveolar ridge
teeth, modified structures
  - teeth, carious
  - tongue, changes
  - macroglossia

OTHERS
  inheritance
  - inheritance, autosomal recessive
supergrroups
  - 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
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 plurimafomalations
cutaneous-genito-neuro-skeletal disorders
dysmorphic face-mental retardation
endocrino-neuro disorders
facio-genito-neuro-skeletal disorders

Super aggreg.
Aggregations: foetal changes, recognized by laboratory data
foetal changes, recognized by ultrasound techniques

LETHALITY
lethality in plurimafomalations, not including chondrodystrophy or chromosomopathies

METABOLIC DISORDERS
carbohydrates, disorders
metabolic acidosis, ketonuria, ketosis, ketogenic, 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 acanthenos 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 chomosomal disorders

Bibliography
OMIM ID: 246200
Smith's Recognizable Patterns of Human Malformation. 5th Edition pag. 599
OMIM ID: 147670
Annuario Orphanet-Italia delle Malattie Rare 2005, pag. 497