| Eponyms: | — Rees syndrome |
| Inheritance: | supposed autosomal recessive  
supposed contiguous genes |
| Semeiological Synthesis: | Neuro-skeletal disorder. Lethal infantile osteopetrosis associated with cerebral atrophy and neuroaxonal spheroids in the CNS and peripheral nerves. May be the same as osteopetrosis-neuronal storage disease. |
| Group | Sub group | Signs: |
| LABORATORY DATA | tissue, biochemical changes  
- neural structures changes |
| MUSCULAR DISEASES | muscular defects, distrectual  
- lethality, in neuromuscular disorders |
| NEUROLOGICAL DISORDERS | brain anomalies  
- brain hypodeveloped, brain atrophy, brain degeneration, encephalopathy, including cerebral palsy  
- corpus callosum, agenesis  
neurological dysfunctions  
- axonal neuropathy |
| PRENATAL-NEONATAL MODIFIED DATA | foetal changes  
- foetal changes, recognized by ultrasound techniques  
prenatal diagnosis  
- prenatal diagnosis, echographic |
| SKELETAL DISORDERS | limb anomalies, limb defects  
- lethality, constant in osteochondrodysplasias  
ossification, changes  
- bone, sclerotic, hyperostotic, not including: focal sclerosis |
| Super group: | neuro-skeletal disorders |
| Super aggreg. | Aggregations: |
| FOETAL CHANGES | foetal changes, recognized by ultrasound techniques |
| LETHALITY | lethality, constant in osteochondrodysplasias  
lethality, in neuromuscular disorders |
| OTHER | axonal neuropathy  
microdeletion syndrome |
| Differential diagnosis: | 4940 ceroid lipofuscinosis, congenital  
19690 osteopetrosis, lethal type  
27482 osteopetrosis-neuronal storage disease  
27914 osteosclerotic bone dysplasia lethal |
| Bibliography | Pediat. Neurosurg. 1995  
OMIM ID: 600329  