

Table 2S. ICD10 codes, total cases, contribution (%) of the registry of RDs, total prevalence in Tuscany (registry of CAs plus registry of RDs) per 100,000 births.

| Group | Rare CA | ICD10 | Total cases | Contribution of the Registry of RDs (%) | Total Prevalence (95%CI) |
|---|---|--------|-------------|---|--------------------------|
| Nervous system | Joubert syndrome | Q04.3 | 1 | 100.0 | 0.39 [0.01-2.17] |
| | Walker-Warburg syndrome | Q04.3 | 2 | 50.0 | 0.78 [0.09-2.82] |
| | De Morsier syndrome (septo-optic dysplasia) | Q04.4 | 1 | 100.0 | 0.39 [0.01-2.17] |
| Eye, hear, face, and neck | Coloboma of optic disc | Q14.2 | 1 | 100.0 | 0.39 [0.01-2.17] |
| Digestive system | Persistent cloaca | Q43.7 | 1 | 100.0 | 0.39 [0.01-2.17] |
| | <i>Alagille syndrome</i> | Q44.71 | 2 | 100.0 | 0.78 [0.09-2.82] |
| Musculoskeletal system | <i>Ehlers-Danlos syndrome</i> | Q79.6 | 1 | 100.0 | 0.39 [0.01-2.17] |
| Skin | <i>Incontinentia pigmenti</i> | Q82.3 | 1 | 0.0 | 0.39 [0.01-2.17] |
| | Hypomelanosis of Ito | | 1 | 0.0 | 0.39 [0.01-2.17] |
| | Ectrodactyly-ectodermal dysplasia-cleft lip/palate (EEC) syndrome | Q82.4 | 1 | 100.0 | 0.39 [0.01-2.17] |
| | Familial dysplastic nevus syndrome | | 1 | 100.0 | 0.39 [0.01-2.17] |
| Phakomatoses | <i>Sturge-Weber syndrome</i> | Q85.81 | 4 | 25.0 | 1.56 [0.43-4.00] |
| Syndromes affecting facial appearance | Antley-Bixler syndrome | Q87.0 | 1 | 100.0 | 0.39 [0.01-2.17] |
| | Freeman-Sheldon syndrome | Q87.0 | 1 | 100.0 | 0.39 [0.01-2.17] |
| | <i>Apert syndrome</i> | Q87.01 | 4 | 50.0 | 1.56 [0.43-4.00] |
| | Moebius syndrome | Q87.06 | 2 | 0.0 | 0.78 [0.09-2.82] |
| Syndromes associated with short stature | Coffin-Siris syndrome | Q87.1 | 1 | 100.0 | 0.39 [0.01-2.17] |
| | Microcephalic osteodysplastic primordial dwarfism (MOPD) | Q87.1 | 1 | 100.0 | 0.39 [0.01-2.17] |
| | <i>Russell-Silver syndrome</i> | Q87.17 | 3 | 33.3 | 1.17 [0.24-3.42] |
| Skeletal | <i>Seckel syndrome</i> | Q87.18 | 1 | 100.0 | 0.39 [0.01-2.17] |
| | Mainzer-Saldino syndrome | Q87.5 | 1 | 100.0 | 0.39 [0.01-2.17] |
| Others | WAGR syndrome | Q87.8 | 2 | 50.0 | 0.78 [0.09-2.82] |
| | Branchio-oto-renal syndrome | Q87.8 | 1 | 0.0 | 0.39 [0.01-2.17] |
| | <i>Williams syndrome</i> | Q87.84 | 4 | 0.0 | 1.56 [0.43-4.00] |
| | <i>Kartagener syndrome</i> | Q89.34 | 2 | 50.0 | 0.78 [0.09-2.82] |
| Chromosomal abnormalities | Wolf-Hirschorn syndrome | Q93.3 | 2 | 50.0 | 0.78 [0.09-2.82] |
| | Gonadal dysgenesis XX | Q99.1 | 1 | 100.0 | 0.39 [0.01-2.17] |
| | <i>Fragile X syndrome</i> | Q99.2 | 2 | 50.0 | 0.78 [0.09-2.82] |

Monogenic rare CAs are reported in italic.

Abbreviations: 95%CI = 95% Confidence Interval; CAs = Congenital Anomalies; RDs = Rare Diseases