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| Σύνδρομο | ORPHAnet code | ICD-10 |
| Rare genetic disease | 98053 | Q87 (*other specified congenital malformation syndromes affecting multiple systems*) |
| Rare chromosomal anomaly | 68335 | Q90-Q99 (*chromosomal abnormalities not elsewhere classified*) |
| Prader-Willy syndrome | 739 | Q87.1 |
| Noonan syndrome | 648 | Q87.1 |
| FRAXF syndrome | 100974 | Q99.2 |
| Di George syndrome | 567 | D82.1 |
| Rett syndrome | 778 | F84.2 |
| Sotos syndrome | 821 | Q87.3 |
| Kabuki Syndrome | 2322 | Q89.8 |
| Williams syndrome | 904 | Q93.82 |
| Down syndrome | 870 | Q90 |
| Angelman syndrome | 72 | Q93.5 |
| Cornelia de Lange | 199 | Q87.1 |