ERN	Diseases	New criteria
Endo-ERN	○ Rare forms of type 1 diabetes ○ Charge syndrome/Ulnar-Mammary syndrome ○ Premature ovarian insufficiency	No
ERN ERNICA	○ Additional esophageal malformations: Eosinophilic Esophagitis; Idiopathic achalasia; Moyamoya disease with early-onset achalasia; Achalasia- alacrimia syndrome. ○ Additional Intestinal malformations/diseases: Intestinal disease due to fat malabsorption (including Abetalipoproteinemia, Acrodermatitis enteropathica, chylomicron retention disease and Idiopathic malabsorption due to bile acid synthesis defects); Sacrococcygeal teratoma ○ Syndromic diaphragmatic or abdominal wall malformations : Familial omphalocele syndrome with facial dysmorphism, Lethal omphalocele-cleft palate syndrome, Omphalocele syndrome Shprintzen-Goldberg type, Omphalocele-diaphragmatic hernia-cardiovascular anomalies-radial ray defect syndrome, pericardial and diaphragmatic defect, diaphragmatic defect-limb deficiency-skull defect syndrome, diaphragmatic hernia-short bowel-asplenia syndrome	
ERN EURACAN	○ Molecular alterations shared by sub-types (NTRK and RET) of breast, colon and lung cancer ○ other rare digestive neoplasia not covered by the present subdomain of domain 5 of EURACAN (e.g. carcinoma of the small bowel) ○ other rare urological neoplasia not covered by the present subdomain of domain 3 of EURACAN (e.g. rare tumours of the kidney)	Y
ERN eUROGEN	○ Rare or complex urogenital reconstructions (highly specialised surgical procedures – Urinary diversion including bladder augmentation, mitrofanof, monti; penile reconstructions and vaginal replacements) ○ Surgery in transgender patients ○ Highly specialised surgical procedures in rare renal cancers (nefrectomy, tumor nefrectomy, tumor nefrectomy with caval embolus)	Y
ERN GENTURIS	○ Paraganglioma, ○ Carney Complex, ○ Hereditary Papillary Renal Carcinoma, ○ Ataxia-Telangiectasia, ○ Bloom syndrome, ○ Nevoid basal cell carcinoma syndrome, ○ Werner Syndrome,	N

	○ Von Hippel Lindaus disease, ○ CMMRD, ○ Gorlin syndrome, ○ Hereditary Leiomyomatosis and Renal Cell Cancer	
ERN LUNG	○ Sarcoidosis; ○ Cystic Lung Diseases; ○ Disorders of the Respiratory Drive; ○	N
ERN RARE-LIVER	○ Genetic cholestatic disease: Alagille ○ Vascular Liver Diseases: Budd-Chiari-Syndrome, non-cirrhotic portal hypertension, vascular malformations ○ Rare causes of acute liver failure ○ Liver disease in pregnancy.	N
ERN Skin	○ Non bullous complex auto immune/inflammatory cutaneous diseases, Premature Skin Ageing, Rare cutaneous proliferation in children and adults	Y
VASCERN	○ Fibromuscular Dysplasia (FMD) ○ Spontaneous coronary artery dissection (SCAD)	Y