
Enfermedades poco frecuentes

Condiciones recomendadas por el Colegio Estadounidense de Obstetricia y Ginecología (ACOG)

Beta-thalassemia – Canavan disease – Cystic fibrosis – Familial dysautonomia – Sickle cell disease – Tay-Sachs disease

Condiciones recomendadas para judíos Ashkenazi

Bloom syndrome – Canavan disease – Cystic fibrosis – Dihydropyrimidine dehydrogenase deficiency – Factor XI deficiency – Familial dysautonomia – Fanconi anemia – Gaucher disease – Glycogen storage disease, type 1A – Maple syrup urine disease – Mucopolidosis IV – Niemann-Pick disease – Tay-Sachs disease – Tyrosinemia – Usher syndrome, type 1F

Condiciones de portación adicionales

21-Hydroxylase-deficient congenital adrenal hyperplasia – 3-Methylcrotonyl-CoA carboxylase deficiency – Achromatopsia – Acrodermatitis enteropathica – Alkaptonuria – Alpha-1 antitrypsin deficiency – Alpha-mannosidosis – Amyotrophic lateral sclerosis – Andermann syndrome – Argininosuccinate lyase deficiency – ARSACS – Aspartylglucosaminuria – Ataxia with vitamin E deficiency – Ataxia-telangiectasia – Autoimmune polyglandular syndrome, type I – Bardet-Biedl syndrome, BBS1-related – Bartter syndrome type 4A – Beta-ketothiolase deficiency – Biotinidase deficiency – Carnitine deficiency, primary systemic – Carnitine palmitoyltransferase II deficiency – Cartilage-hair hypoplasia – Cerebrotendinous xanthomatosis – Choroideremia – Citrullinemia, type I – Cohen syndrome – Combined pituitary hormone deficiency, PROP1-related – Congenital disorder of glycosylation type Ia – Costeff optic atrophy syndrome – Crigler-Najjar syndrome – Cystinosis – Diabetes, permanent neonatal – Dubin-Johnson syndrome – Ehlers-Danlos syndrome, dermatosparaxis – Ashkenazi Jewish Conditions – Bloom syndrome – Canavan disease – Cystic fibrosis – Dihydropyrimidine dehydrogenase deficiency – Factor XI deficiency – Familial dysautonomia – Fanconi anemia – Ehlers-Danlos syndrome, hypermobility – Ehlers-Danlos syndrome, kyphoscoliotic – Factor V Leiden thrombophilia – Familial Mediterranean fever – Galactokinase deficiency – Galactosemia – Glutaric acidemia, type 1 – Glycogen storage disease, type Ib – Glycogen storage disease, type III – Glycogen storage disease, type V – GM1-gangliosidosis – Hearing loss, DFNB1 and DFNB9 nonsyndromic – Hearing loss, DFNB59 nonsyndromic – Hemochromatosis – Hemoglobin C – Hemoglobin D – Hemoglobin E – Hemoglobin O – Hereditary Fructose Intolerance – Herlitz junctional epidermolysis bullosa, LAMA3-related – Herlitz junctional epidermolysis bullosa, LAMB3-related – Herlitz junctional epidermolysis bullosa, LAMC2-related – HMG-CoA lyase deficiency – Homocystinuria, cblE type – Homocystinuria, classic – Hurler syndrome – Hypophosphatasia, autosomal recessive – Inclusion Body Myopathy 2 – Juvenile retinoschisis, X-linked – Krabbe disease – Lipoamide dehydrogenase deficiency – Lipoprotein lipase deficiency, familial – Medium-chain acyl-CoA dehydrogenase deficiency – Megalencephalic leukoencephalopathy with subcortical cysts – Gaucher disease – Glycogen storage disease, type 1A – Maple syrup urine disease – Mucopolidosis IV – Niemann-Pick disease – Tay-Sachs disease – Tyrosinemia – Usher syndrome, type 1F – Methylmalonic acidemia – Mucopolidosis II – Mucopolidosis III – Multiple carboxylase deficiency – Nephrotic syndrome, steroid-resistant – Neuronal ceroid lipofuscinosis, CLN3-related – Neuronal ceroid lipofuscinosis, CLN5-related – Neuronal ceroid lipofuscinosis, CLN8-related – Neuronal ceroid lipofuscinosis, PPT1-related – Neuronal ceroid lipofuscinosis, TPP1-related – Nijmegen breakage syndrome – Pendred syndrome – Phenylketonuria – Polycystic kidney disease – Pompe disease – Prekallikrein deficiency – Primary hyperoxaluria, type 1 – Primary hyperoxaluria, type 2 – Primary hyperoxaluria, type 3 – Propionic acidemia – Prothrombin deficiency – Rhizomelic chondrodysplasia punctate type 1 – Rh-null syndrome – Rickets, pseudovitamin D-deficiency – Salla disease – Sandhoff disease – Short-chain acyl-CoA dehydrogenase deficiency – Sick sinus syndrome – Smith-Lemli-Opitz Syndrome – Spherocytosis, hereditary – Tay-Sachs pseudodeficiency – Thrombocytopenia, congenital amegakaryocytic – Tyrosine Hydroxylase Deficiency – Very long-chain acyl-CoA dehydrogenase deficiency – Von Willebrand disease type 2 Normandy – Von Willebrand disease type 3 – Wilson disease – Zellweger syndrome spectrum, PEX1-related – Metachromatic leukodystrophy