Molekulární diagnostika vrozených nemocí

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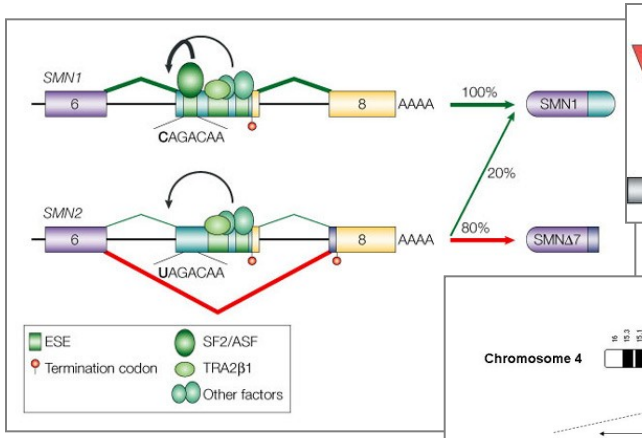
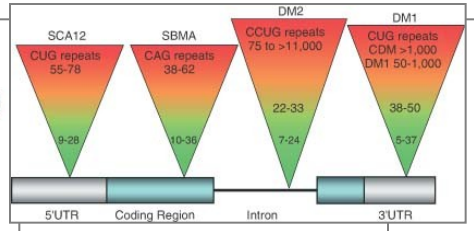
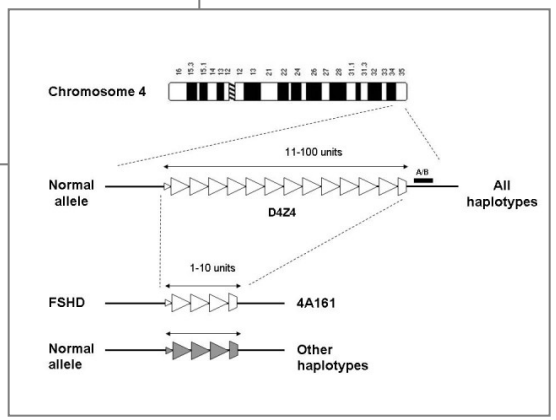
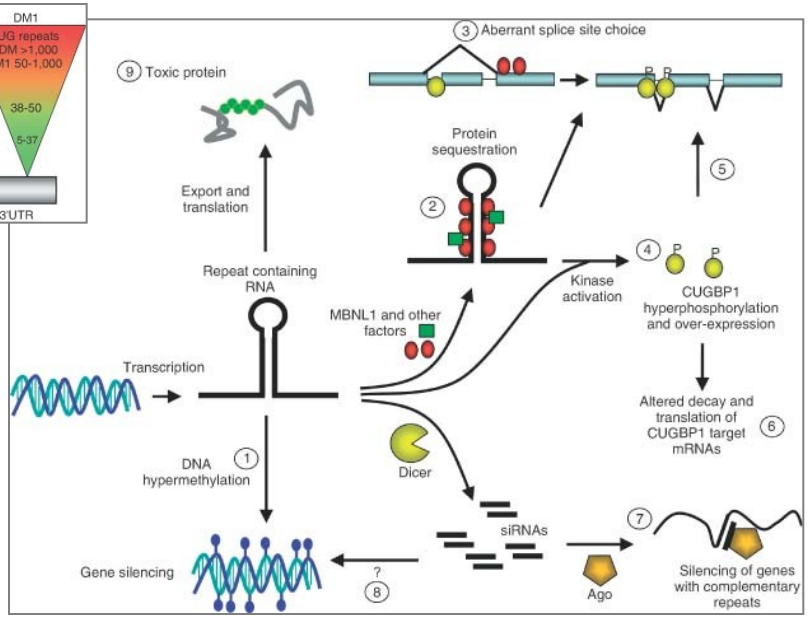
Molekulární problematika dědičných

- nervosvalových nemocí
- metabolických nemocí
- mentálních retardací
- kožních nemocí



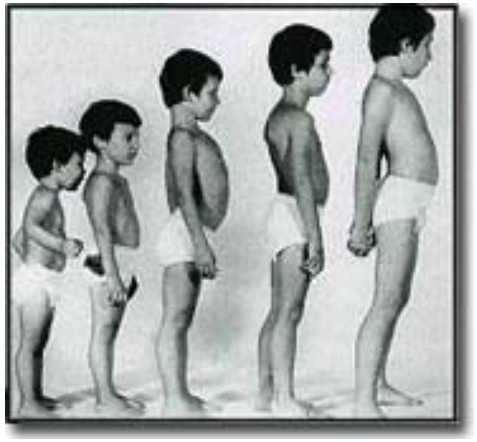


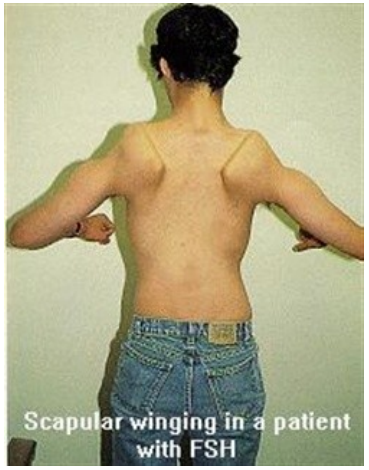




Nervosvalové nemoci

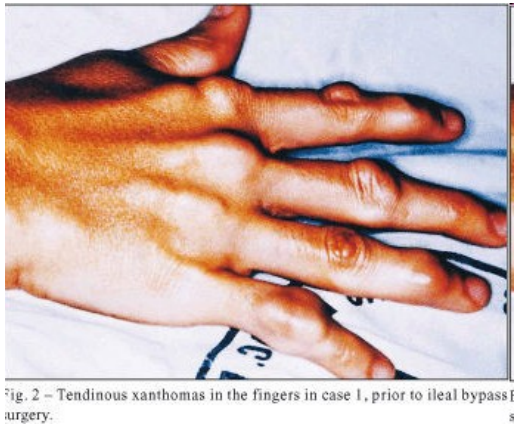
Disease	*Gene*	*Localization*	*Protein*
Duchenne/Becker muscular dystrophy	*DMD*	Xp21	Dystrophin
Spinal muscular atrophy	*SMN1*	5q12	Survival motor neuron protein 1
Myotonic dystrophy, type 1	*DMPK*	19q13	Dystrophia myotonia protein kinase
Myotonic dystrophy, type 2	*ZNF9*	3q13	Zinc finger protein 9
Facioscapulohumeral muscular dystrophy	Deletion of D4Z4	4q35	
Myotonia congenita, Thomsen/Becker	*CLCN1*	7q35	Chloride channel 1
Myotonia congenita	*SCN4A*	17q23	Sodium channel 4, alpha subunit
Limb girdle muscular dystrophy, type 2A	*CAPN3*	15q15	Calpain-3
Limb girdle muscular dystrophy, type 2D	*SGCA*	17q12	Sarcoglycan, alpha
Limb girdle muscular dystrophy, type 2I	*FKRP*	19q13	Fukutin-related protein



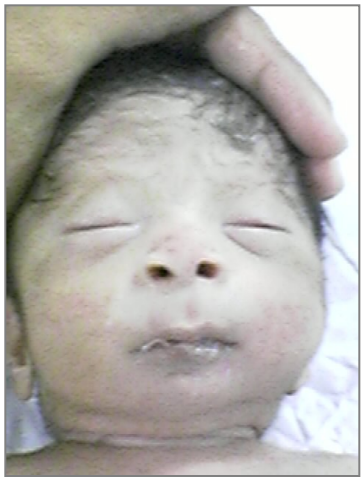


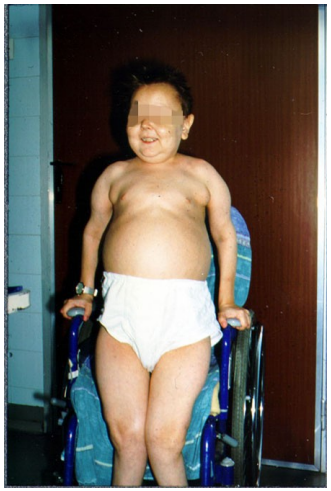
Metabolické nemoci

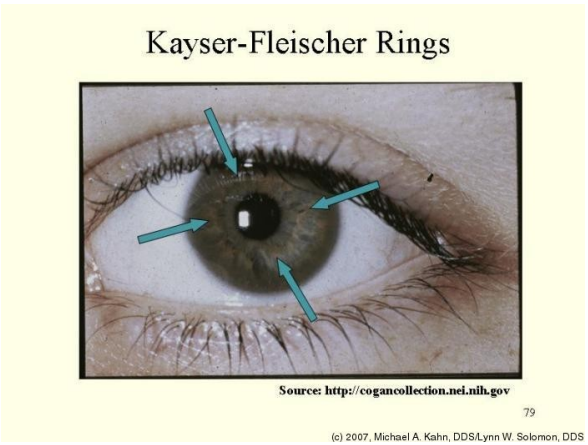
Disease	*Gene*	*Localization*	*Protein*
Familial hypercholesterolemia	*LDLR*	19p13	Low density lipoprotein receptor
Familial hypercholesterolemia, type B	*APOB*	2p24	Apolipoprotein B100
Alpha-1-antitrypsin deficiency	*PI*	14q32	Alpha-1-antitrypsin
Phenylketonuria	*PAH*	12q24	Phenylalanine hydroxylase
Smith-Lemli-Opitz syndrome	*DHCR7*	11q12	Sterol delta-7-reductase
Wilson disease	*ATP7B*	13q14	ATPase, Cu(2+)-transporting beta polypeptide
Congenital adrenal hyperplasia	*CYP21A2*	6p21	21-hydroxylase
Galactosemia	*GALT*	9p13	Galactose-1-phosphate uridylyltransferase
Glycogen storage disease, type 1A	*G6PC*	17q21	Glucose-6-phosphatase
Glycogen storage disease, type 1B	*G6PT*	11q23	Glucose-6-phosphate transporter protein
Glycogen storage disease, type 2	*GAA*	17q25	Acid alpha-1,4-glucosidase
Glycogen storage disease, type 3	*AGL*	1p21	Glycogen debrancher enzyme



ig. 2 – Tendinous xanthomas in the fingers in case 1, prior to ileal bypass
urgery.

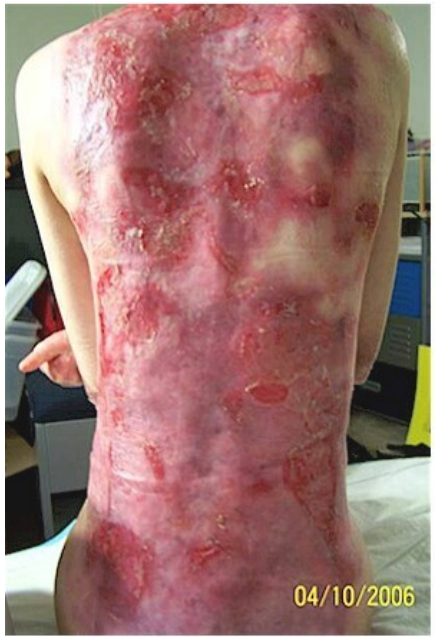


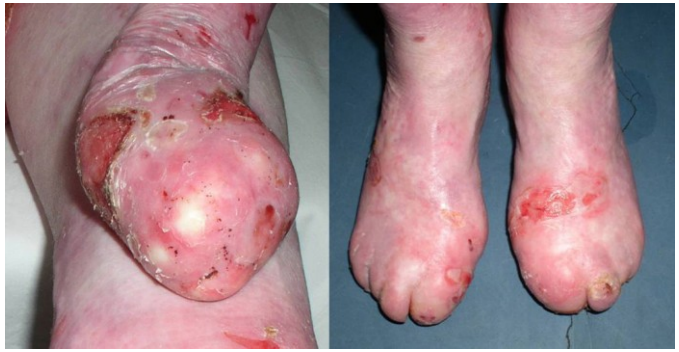


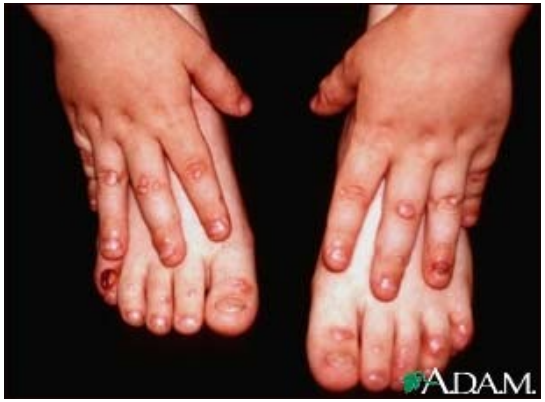


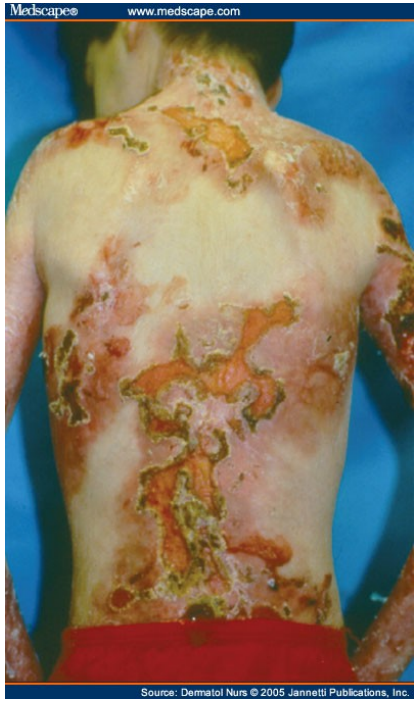
Kožní nemoci

Disease	*Gene*	*Localization*	*Protein, function*
Epidermolysis bullosa simplex	*KRT5, KRT14*	12q13, 17q12	Keratin 5, keratin 14
Epidermolysis bullosa dystrophica	*COL7A1*	3p21	Type VII collagen, alpha-1 chain
Incontinentia pigmenti	*NEMO* (*IKBKG*)	Xq28	
Ichthyoses	*FLG, STS*, *TGM1*	1q21, 14q12, Xp22	Filaggrin, steroid sulfatase, transglutaminase,



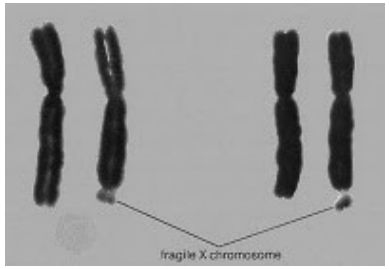


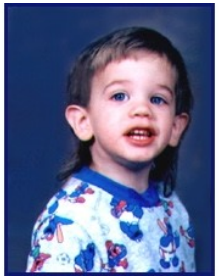




Mentální retardace

Disease	*Gene*	*Localization*	*Protein, function*
Fragile X syndrome	*FMR1*	Xq27	Fragile X mental retardation protein





Epilepsie

Disease	*Gene*	*Localization*	*Protein, function*
GEFS+	*SCN1A*	2q24	Sodium chanel, neuronal type I, alpha subunit
pyridoxine-dependent epilepsy, pyridoxamine phosphate oxidase deficiency, hyperprolinémia	*ALDH7A1, PNPO, ALDH4A1*	5q31; 17q21; 1q36	Aldehyde dehydrogenase 7 family, memner A1; Pyridoxamine 5-prime-phosphate oxidase; Aldehyde dehydrogenase, family 4, subfamily A

Onemocnění ….. asociovaný gen ….. dědičnost (AR, AD, X-vázaná) ….. kódovaný protein ….. funkce proteinu ….. vliv mutace/mutací na funkci proteinu ….. molekulární podstata onemocnění ….. klinické projevy onemocnění ….. používané metody DNA diagnostiky