

**LANGE BIOCHEMISTRY AND GENETICS FLASH CARDS  
2/E (LANGE FLASHCARDS)**

Louis Fiorillo

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NPD type A presents in the first 6 months of life with rapidly progressive CNS deterioration seizurespasticity, and failure to thrive. Purine degradation takes place in most tissues.

BiochemistryandGeneticswillbeoneofthemostpowerfultoolstohelpprepare Skeletal muscle, brain, kidney, and liver. On further questioning, the patient does not eat yogurt, ice cream, or any other dairy products throughout the day. You tell the parents that you believe that their son is suffering from an autosomal recessive disorder caused by defects in two lysosomal enzymes.

OnlysomepatientswithtypeIIdiseasewillhavemacularcherry-redspots. The symptoms include hepatosple- nomegaly, corneal clouding, large tongue, developmental delay, dwarfism, hearing loss, and mental retardation.