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S-2-5
GENE DIAGNOSIS AND THERAPY OF NEONATAL INHERITED METABOLIC DISEASES

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Characterization of mutations responsible for human diseases is accelerating by the completion of the Human Genome Project. Gene diagnosis for an inherited disease mostly involves the detection of mutation of the corresponding gene, which require prior knowledge of the mutations commonly found in a population. Cystic fibrosis and medium chain acyl-CoA dehydrogenase (MCAD) are examples with this aspect in the Caucasian. In Oriental populations, however, this is not the case. The tetrahydrobiopterin (BH4) deficient hyperphenylalaninemia (HPA) caused by 6-pyruvoyltetrahydropterin synthase (PTS) deficiency and glucose-6-phosphate dehydrogenase (G6PD) deficiency offer a good illustration. Our previous studies indicated that the mutations harbored in the PTS and G6PD genes were confined to certain common mutations in Chinese populations (PTS: 155A>G, 259C>T, 286G>A; G6PD: 95A>G, 1376G>T, 1388G>A), which were different from other populations. These should help a more precise diagnosis when a newborn is suspected by newborn screening or clinical symptoms and therefore offer the appropriate treatment in time. General treatments for inherited metabolic disorders often involve substrate restriction, replacement of deficient products, toxic metabolites removal, and avoidance of symptom inducing agents. Enzyme replacement, gene therapy and stem cell transplantation are currently under research and may be applied to treat these diseases in the future.

S-3-1
PREANALYTICAL INFLUENCES ON PHYSIOLOGICAL VALUES IN THE ELDERLY

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Once collected, specimens from the elderly are influenced by all the same factors that affect specimens drawn from any other patient population. However, the sometimes greater difficulty in performing venipunctures may lead to a higher incidence of hemolysis. The elderly are subject to most of the preanalytical variables that affect younger adults but they may respond differently from younger individuals to the administration of drugs because of changes in both hepatic and renal function. Such changes may be compounded by subclinical illness, such as hypertension or anemia. The basal metabolic rate in the elderly is decreased, probably associated also with reduced food intake which, in turn, affects the concentration of plasma proteins. Old people tend to have reduced muscle mass so that those tests whose results are associated with muscle and skeletal mass are affected by age. The concentrations of many hormones are affected by aging: some exhibit dramatic reductions while others show marked increases. The sensitivity of target organs to hormones tends to be reduced. Circadian variation of hormone concentrations may be altered in the elderly. The differences between sexes of concentrations of analytes that are affected by hormones lessen considerably in the elderly.

S-3-2
LABORATORY MARKERS OF AGING: THE EXAMPLE OF HORMONES

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Physiological changes associated with aging influence the reference intervals of hormones in the elderly. These changes may be manifold ranging from alterations in the homeostatic control by the hypothalamus-pituitary-adrenal axis to neuronal changes, and dysregulation of cytokine production such as an increase in interleukin-6 (IL-6) with age. Gender related differences need also to be kept in perspective. Thus unconjugated dehydroepiandrosterone (DHEA) levels are higher in women when compared to men. Some examples of hormones that increase with age are Follicle-stimulating hormone (FSH), Luteinizing hormone (LH), Cortisol and antidiuretic hormone. Indeed the levels of antidiuretic hormone in healthy subjects ranging from 53 to 87 years can be twice as high as seen in the 21 to 51 year age group. Some examples of hormones that decrease with age are testosterone, dopamine, serotonin, aldosterone, growth hormone. A large population-based study of men aged 40 to 70 has documented decreases in both total and free testosterone with increasing age. Discrepancies in literature on reference intervals of hormones in the elderly may be due to choice of healthy subjects with an overlooked nutritional trace element deficiency.