

## Genetic Counseling and Education

## AB127. Provision of medical genetic services in Thailand: Siriraj experience

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**Background:** Genetic services at the Department of Pediatrics, Faculty of Medicine Siriraj Hospital was established since 1960, providing genetic counseling and cytogenetic laboratory. Since 1987, the clinical genetic services have expanded to include other areas with a mandate of providing for comprehensive genetic services with a primary role in diagnosis, counseling and prevention.

**Methods:** The establishment of genetic services at Siriraj Hospital was reviewed.

**Results:** Our services include (I) Genetic counseling program: 4 specialty clinics Genetic Clinic received referrals from Bangkok and provincial hospitals all over the country; Birth Defects Clinic (established in 1991) providing services for patients with cleft lip/palate, neural tube defects, arthrogyposis, and limb anomalies; Genetic Skeletal Dysplasia Clinic [1992] providing services for patients with achondroplasia, osteogenesis imperfecta, and metabolic bone diseases (mucopolysaccharidosis); Down Syndrome Clinic [1993] serving more than 600 families, providing genetic counseling, medical checkup, referral

to early stimulation program and assisting these children toward integration; (II) Genetic screening program: newborn screening for genetic metabolic disorders (congenital hypothyroidism and phenylketonuria) has been a routine screen since year 2005. We have provided expanded newborn screening to detect 40 disorders of inborn errors of metabolism (IEM) since 2014, and have performed the screening more than 35,000 samples per year; (III) Genetic laboratory: initially only conventional cytogenetic analysis, subsequently biochemical genetics laboratory was established. Currently numerous IEM e.g., amino acid, carbohydrate, urea cycle, organic acid, fatty acid oxidation are being identified. The Genetic Metabolic Center was established since June 2001 and has served infant and children with IEM all over the country; (IV) non-governmental organization: Down Syndrome Parents Support Group at Siriraj Hospital was founded in 1993, providing more holistic approach and personal services to individuals and their families.

**Conclusions:** Genetic services at Siriraj may serve as a model for public health setting.

**Keywords:** Down syndrome; expanded newborn screening; genetic counseling

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