AB124. Craniofacial anomalies: an experience at Siriraj Hospital

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Background: Craniofacial anomalies (CFA) result from interruption of normal embryologic growth and differentiation of the face and skull, and can be classified in several groups, such as branchial arch syndromes, craniosynostosis, orofacial clefts, and encephalocele. Management of CFA requires multidisciplinary team. Cranio-maxillo-facial clinic at Siriraj hospital was founded more than 15 years, and consists of experts from multiple medical and surgical specialties, such as plastic surgeon, neurosurgeon, ophthalmologist, otolaryngologist, orthodontist, and clinical geneticist.

Methods: A retrospective chart review of all children clinically diagnosed with CFA who were seen at Siriraj hospital was performed. Specific syndromes in which CFA are the primary feature will be reviewed here.

Results: Common syndromes with CFA in our hospital are orofacial clefts, especially cleft lip/palate, and oculo-auriculo-vertebral (OAV) spectrum. In recent years, we found that diabetic embryopathy and prenatal exposure to teratogen increase in patients with CFA.

Conclusions: Clinical approach in diagnosing syndromes associated with CFA includes good history taking (prenatal, perinatal and postnatal history), family history, careful physical examination, and investigations. Clinical diagnosis may require gestalt diagnosis and pattern recognition on several key features. However, although advances in genetic testing (e.g., fluorescent *in situ* hybridization, chromosome microarray, whole exome sequencing) play an important role in diagnosis of genetic syndromes associated with CFA; many genetic syndromes still require the expertise of clinical geneticist in clinical recognition which is regard as the cornerstone of establishing the diagnosis.

Keywords: Craniofacial anomalies (CFA); craniosynostosis; orofacial cleft; oculo-auriculo-vertebral spectrum (OAV)

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