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Midline facial defects with associated brain abnomality

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A newborn girl weighing 2,700 g, born at 37 weeks 2 days of gestation to a 40-year-old multigravida mother via vaginal delivery was admitted to neonatal intensive care unit with cyanosis and morphologic anomalies. On physical examination, she showed hypotonia, microcephaly, low set ears, and midline facial defects such as single naris, depressed nasal ridge, and cleft lip (Figure 1). Brain MRI (magnetic resonance imaging) showed alobar holoprosencephaly with fused cerebral hemispheres and the thalamus shaped like a heart (Figure 2).

Chromosomal analysis of her peripheral blood revealed 46, XX, del (18) (p11.1). Holoprosencephaly is a rare malformation of the human forebrain. This anomaly occurs due to failed cleavage of the prosencephalon early in gestation[1,2]. Classically three subtypes have been recognized. The three main subtypes, in order of decreasing severity are alobar, semilobar, and lobar holoprosencephaly. It may be associated with other chromosomal anomalies such as trisomy 13, 18, chromosome 7q, 2q, or 18p deletion[3].

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Figure 1. A 1-month-old girl with low set ears, and midline facial defects such as single naris, depressed nasal ridge, and median cleft lip.

Figure 2. T1-weighted coronal brain MRI shows alobar holoprosencephaly with fused cerebral hemispheres and the thalamus shaped like a heart

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환자명(Print name): 양혜승 (Hyeo Seung Yang)

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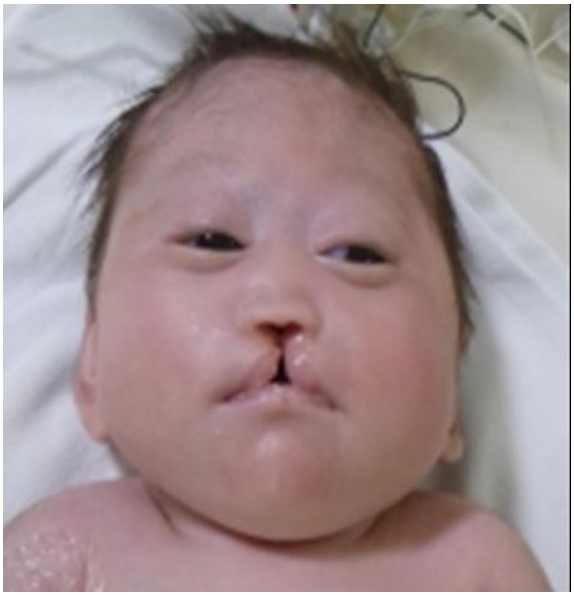

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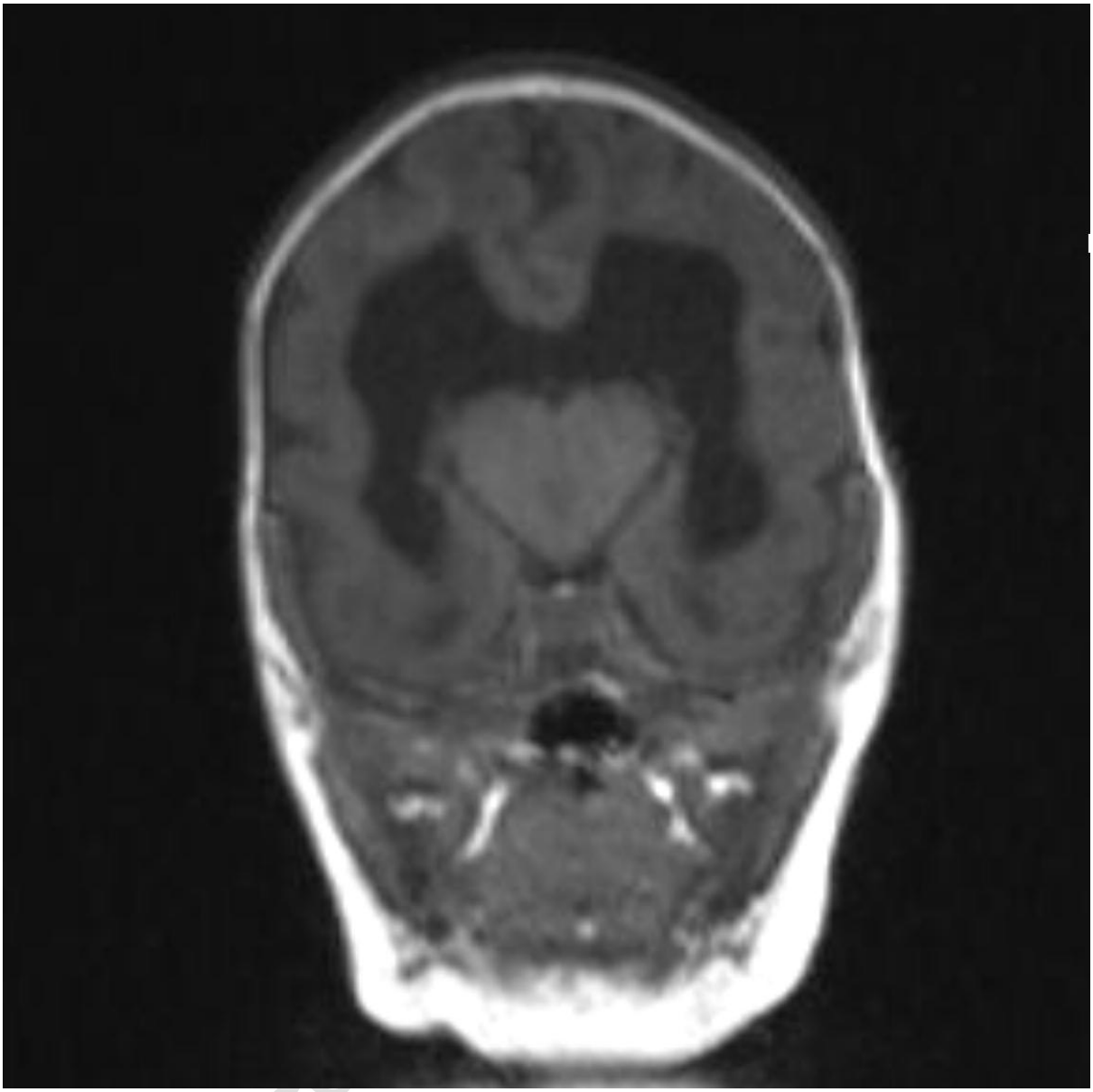

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