Listen to Your Patients: A Diagnostic Clue

A newborn female child was delivered by cesarean after 37 weeks of gestation, complicated by intrauterine growth retardation. Her weight was 1610 g (<3rd percentile), length 42 cm (<3rd percentile), and occipitofrontal circumference 29 cm (<3rd percentile). She was noted to have micrognathia and mild hypotonia. An echocardiogram showed multiple, small ventricular septal defects; no abnormalities were detected by cerebral and abdominal ultrasound examinations. In the first days of life, she was noted to have a high-pitched, monochromatic cry, resembling the sound of a cat [**Audio File**; available at www.jpeds.com]. Cri-du-chat syndrome (OMIM #123450) was suspected and a karyotype with banding confirmed the terminal deletion of a portion of the short arm of chromosome 5 (**Figure**).

Cri-du-chat syndrome, also known as 5p deletion syndrome, or 5p- syndrome, was first described in 1963 by Lejeune et al[1] and is among the most common chromosomal abnormalities with an incidence ranging from 1 in 15 000 to 50 000 live births.[2,3] Clinical features are variable and often include microcephaly, round face, hypertelorism, epicanthal folds, micrognathia, low-set ears, hypotonia, developmental delay, intellectual disability, and malformations of brain, heart, and genitourinary system. Although many affected individuals have similar overlapping deletions, there is no common recurring breakpoint. The high pitched cry has been attributed to multiple regions within the short arm of chromosome 5 and likely depends on congenital anomalies of a narrow and diamond-shaped larynx along with a floppy and small epiglottis.[2,4]

Physicians have always used their bodies as an instrument to make a diagnosis: inspection, palpation, percussion, and auscultation, and even smell and hearing. Even with highly technological diagnostic advances, a doctor's senses have not been yet replaced by instruments and tests, as illustrated by this case.■

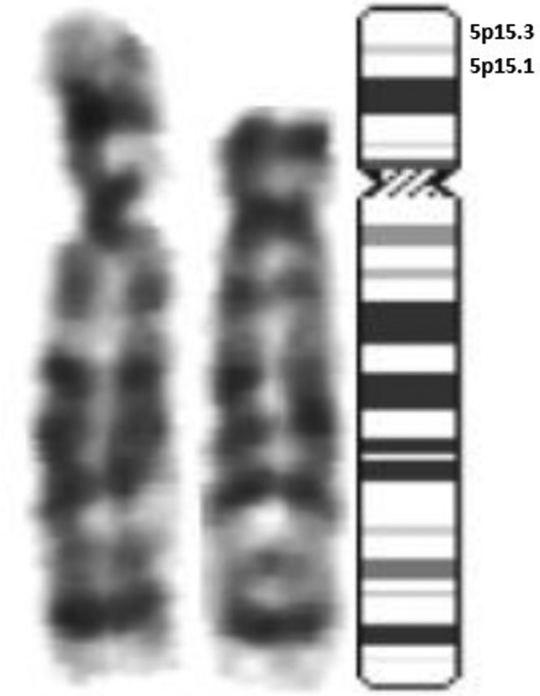

Figure. Partial chromosome analysis of the presented case showing a terminal deletion of a portion of the short arm of chromosome 5.

The authors declare no conflicts of interest.

Data Statement

Data sharing statement available at www.jpeds.com.

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