

Monosomy 18p and hepatosplenomegaly

Kemawikasit Ponchai, Evelyn Erickson, Eduardo A Marrero

To the Editors,

An 11-month-old infant, previously known to have a chromosome 18p- deletion, came to the clinic for her six months old well child care visit. Palpable hepatosplenomegaly was found and as a result, an abdominal ultrasound and complete blood count (CBC) were ordered. The CBC was normal except for monocytosis of $1.7 \times 10^3/\text{mm}^3$ (normal- $0.1-1 \times 10^3/\text{mm}^3$). The abdominal ultrasound confirmed hepatosplenomegaly of 9.2 cm found on physical examination. As per her mother, three months prior to her visit, while traveling abroad, the infant had an episode of clear nasal discharge, wet cough, intermittent fever for 3–4 days and watery diarrhea.

The patient was born to a 45-year-old mother, full term at 38 weeks with intrauterine growth restriction and a birth weight of 1970 g. She was born by C-section due to a category 2 tracing. Her mother had chronic hypertension and was on methyldopa. There were no signs of chorioamnionitis. The baby was admitted to the Neonatal Intensive Care Unit for eight days due to low birth weight. The quad screen test showed increased risk of neural tube defects and a fetal amniocentesis reported 46 XX del (18)(p10). Meanwhile, the FISH was that of a

normal female. The geneticist confirmed the results of the amniocentesis, and suggested maternal testing after proper family education and genetics counseling.

The patient's weight ranged from below 5–10% in weight, and the height was below 5%. She was able to sit unsupported but was not able to pivot when sitting, did not crawl well, and did not cruise. She held her bottle and used an immature pincer grasp, did not say 'mama, dada' and did not play gesture games. On the physical examination height was 65 cm (<5%), weight 7.3 kg (<5%) and head circumference 45 cm (75%) (Figure 1). The peripheral oxygen saturation was 95% and her heart rate 155 beats per minute. She was alert, active, well hydrated and not in acute distress. Her eyes, ears, nose and throat appeared normal. She had a short webbed neck. The heart and chest were normal although her right lung had mild wheezing in the lower lung field. Her abdomen was positive for a hepatomegaly and a splenomegaly. No costovertebral angle tenderness was observed.

Among the most important physical findings in patients with monosomy 18p- are ventricular septum defect, patent ductus arteriosus, inguinal hernia,

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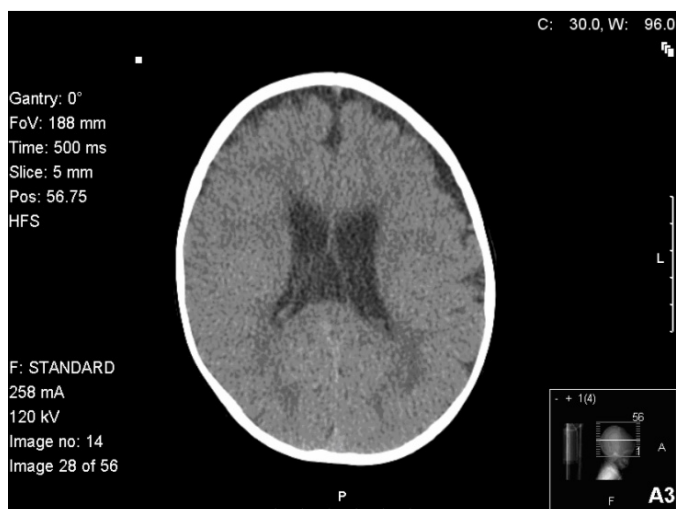


Figure 1: Computed tomography of the brain taken without contrast. Patient had this tomography in order to rule out brain calcifications as well as congenital brain anomalies.

accessory spleen, orthopedic anomalies, pes planus, scoliosis or kyphosis, spina bifida occulta, neurological anomalies (hypotonia, dystonia, holoprosencephaly, agenesis of corpus callosum), strabismus, ptosis, cleft lip/palate, growth hormone deficiency, recurrent otitis media and hearing loss. More than 2/3rd patients have developmental delays, speech delays and/or mild mental retardation [1]. Some can also have psychiatric disturbances such as attention deficit hyperactivity disorder, anxiety, mania, depression and psychosis [2]. Hepatosplenomegaly has not been documented to be associated with monosomy 18p.

Further evaluation was needed to evaluate whether the hepatosplenomegaly found in this patient was linked to monosomy 18p or not. Antibodies for cytomegalovirus (CMV) IgG was found elevated in this patient (Index Value: 4.99, Normal: 0–0.90). A CT of the head (Figure 1) was done which ruled out periventricular calcifications, pituitary abnormalities, mild form of holoprosencephaly with missing corpus callosum, although the patient reported chronic suppurative otitis media of the left ear. An ophthalmology consult was done and found to be within normal limits. In subsequent visits, the liver and the spleen decreased in size. No association between monosomy 18p- and hepatosplenomegaly was found. We concluded that her visceromegaly was related to a past CMV infection. It is still important in patients with monosomy 18p to be closely followed when medical illnesses arise to avoid any medical complications [3].

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Author Contributions

Kemawikasit Ponchai – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

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Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

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