Clin. Lab. 2023;69:1-5

CASE REPORT

Phenotypic Spectrum of Trisomy 18 Mosaicism: a New Patient and Literature Review

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SUMMARY

Background: **Trisomy 18 syndrome, also called Edwards syndrome, is the second most common autosomal trisomy after trisomy 21 that is caused by the presence of an extra copy of chromosome 18. Approximately 50% of infants with trisomy 18 cannot survive for more than 1 week and about 5 - 10% of children die within 1 year after birth. The aim of this study is to describe a 4-year-old female patient of mosaic trisomy 18 with normal prenatal ultrasound findings and maternal serum markers and to investigate the relationship between the percentage of trisomic cells and the major clinical phenotypes combined with other nine patients through a review of the literature.**
Methods: **The patient's peripheral blood was examined by cytogenetic G-banding technique.**
Results: **The cytogenetics results reported following the ISCN 2020 guideline as mos 47,XX,+18[87]/46,XX[13].**
Conclusions: **There is little correlation between various phenotypes of mosaic trisomy 18 and the percentage of trisomy cells in the patient's peripheral leukocytes. Although most of fetuses with mosaic trisomy 18 have abnormal ultrasound findings, it is necessary to highlight the possibility of normal findings during the pregnancy.**
(Clin. Lab. 2023;69:xx-xx. DOI: 10.7754/Clin.Lab.2022.220610)

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Case Report accepted June 13, 2022

Supplementary Data

Table S1. Phenotypic spectrum of trisomy 18 mosaicism.

	This case	A. Perrin, 2009	Siddharth Banka, 2011 CASE 1	Siddharth Banka, 2011 CASE 2	Elaine Maria Pereira, 2012	Ana Laura Fitas, 2013	Serena Motta, 2015	Neidin Bussmann 2015 F1	Neidin Bussmann 2015 F2	Naveed Ahmad 2016
Gender	**Femal**	**Male**	**Femal**	**Male**	**Femal**	**Femal**	**Male**	**Femal**	**Femal**	**Male**
Age (years)	**4**	**31**	**11**	**5**	**10**	**5 months**	**17**			**18 months**
Gestation (weeks)	**41**		**37**	**38**	**32**	**41**	**37**	**29 + 5**	**29 + 5**	**38**
Cytogenetic Findings										
Karyotype (blood)	**47,XX, +18[87]/ 46,XX [13]**	**47,XY, +18[13]/ 46,XY [16]**	**47,XX, +18[44]/ 46,XX [6]**	**47,XY, +18[12]/ 46,XY [18]**	**47,XX, +18[60]/ 46,XX [140]**	**47,XX, +18[26]/ 46,XX [3]**	**47,XY, +18[10]/ 46,XY [15]**	**47,XX, +18[8]/ 46,XX [42]**	**47,XX, +18[4]/ 46,XX [46]**	**47,XY, +18[6]/ 46,XY [24]**
Karyotype (skin)		**3%**	**47,XX, +18[15]/ 46,XX [35]**			**47,XX, +18[5]/ 46,XX [24]**	**47,XY, +18[5]/ 46,XY [95]**	**46,XX**		
Karyotype (buccal smear)		**L-65%, R-50%**						**46,XX**	**47,XX, +18[51]/ 46,XX [35]**	
Karyotype (tumor)					**46,XX**					**47,XY, +18[22]/ 46,XY [23]**
Karyotype (bone marrow)							**47,XY, +18[4]/ 46,XY [19]**			
Prenatal manifestations										
Intrauterine growth retardation	**no**		**yes**	**yes**	**yes**	**no**	**yes**			
Decreased fetal movements	**no**				**yes**	**no**				
Polyhydramnios	**no**		**yes**			**no**				
Growth										
Birth length - cm (% tile)						**46 (normal)**				
Birth weight - g (% tile)	**2,550 (< 3%)**		**2,030 (< 3%)**	**1,850 (< 3%)**	**680 (< 3%)**	**2,650 (< 10%)**	**1,680 (< 3%)**	**1,250 (< 10%)**	**1,090 (< 10%)**	**1,800 (< 3%)**
Birth OFC - cm (% tile)			**34,5**	**31.5 (0.4%)**						
Height at evaluation - cm (% tile)	**90 (< 3%)**		**9%**			**57 (< 3%)**				
Weight at evaluation - g (% tile)			**25%**			**3,850 (< 3%)**				
OFC at evaluation - cm (% tile)	**45.3 (< 3%)**		**50% - 75%**	**0,40%**						

Table S1. Phenotypic spectrum of trisomy 18 mosaicism (continued).

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Gender	**Femal**	**Male**	**Femal**	**Male**	**Femal**	**Femal**	**Male**	**Femal**	**Femal**	**Male**
Head & Neck										
Dolichocephaly						yes				
Microcephaly	yes								yes	
Narrow bifrontal diameter						yes				
Frontal bossing	yes					yes				
Prominent occiput			yes			yes				
Large anterior fontanelle									yes	
Bushy eyebrows	yes			yes						
Long eyelashes				yes						
Upslanting palpebral fissures				yes						
Downslanting palpebral fissures						yes				
Downward oblique eye orientation					yes					
Ptosis					yes					
Microphthalmia									yes	
Narrow palpebral fissure									yes	
Low-set ears					left	yes				
Deformed ears	yes				right					
Dysplastic ears								yes		
Retrognathia						yes				
High-arched palate						yes				
Micrognathia									yes	
Neurologic										
Developmental delay	yes			yes	yes		yes			yes
Speech delay	yes			yes						yes
Febrile seizures							yes			
Cardiac										
Ventricular septal defect			yes	yes		yes	yes	yes	yes	yes
Atrial septal defect	yes			yes		yes		yes	yes	
Patent ductus arteriosus				yes				yes	yes	yes
Pulmonary artery stenosis						yes				
Bicuspid aortic valve						yes				

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Gender	**Femal**	**Male**	**Femal**	**Male**	**Femal**	**Femal**	**Male**	**Femal**	**Femal**	**Male**
Hypertrophic cardiomyopathy				yes						
Gastrointestinal										
Tracheoesophageal fistula			yes					yes		
Necrotising enterocolitis										
Constipation			yes							
Vomiting						yes				
Feeding difficulties	yes			yes	yes	yes				yes
Respiratory										
Recurrent respiratory infections	yes		yes			yes	yes			
Dyspnea						yes				
Asthma			yes							
Genitourinary										
Horseshoe kidney			yes							
Micropenis									yes	
Oligoasthenoteratozoospermia		yes								
Skeletal/Muscular Findings										
Short sternum			yes							
Joint laxity	yes			yes						
Scoliosis	yes				yes					
Hypotonia	yes			yes	yes	yes				
Arms/Hands										
Flexed fingers			yes							
Adducted thumbs			yes							
Distal phalangeal crease hypoplasia	yes				yes					
Fifth digit camptodactyly					yes					
Clenched hands									yes	
Single transverse palmar creases									yes	
Legs/feet										
Hypoplastic nails			yes							
Short dorsiflexed great toes			yes							
Overriding toes				yes						
Syndactyly			yes							
Rocker bottom feet	no			yes	right	yes				
Restricted motion of the hip					yes					

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	This case	A. Per-rin, 2009	Siddharth Banka, 2011 CASE 1	Sidd-harth Banka, 2011 CASE 2	Elaine Maria Pereira, 2012	Ana Laura Fitas, 2013	Serena Motta, 2015	Neidin Buss-mann 2015 F1	Neidin Buss-mann 2015 F2	Naveed Ahmad 2016
Gender	**Femal**	**Male**	**Femal**	**Male**	**Femal**	**Femal**	**Male**	**Femal**	**Femal**	**Male**
Asymmetry										
Facial asymmetry			**yes**							**yes**
Hands asymmetry			**yes**							
Kidney asymmetry				**yes**						
Hemihypotrophy						**yes**				
Hemihypertrophy										**yes**
Others										
Inguinal hernia				**yes**				**yes**		**yes**
Skin pigmentation	**following Blaschko's lines**		**following Blaschko's lines**	**yes**	**following Blaschko's lines**	**following Blaschko's lines**				
Tumor					**hepato-blastoma**		**Hodgkin lym-phoma**			**hepato-blasto-ma**