Jarcho Levin Sendromlu Hastalarda Santral Sinir Sistemi veya Nöral Tüp Defekti Birlikteliği; Tek Merkez Tecrübesi

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VAN

JARCHO-LEVİN SENDROMU

- Jarcho-Levin Sendromu (JLS) kosta ve vertebra anomalilerinin görüldüğü, nöral tüp defekti, Arnold Chiari malformasyonu, renal ve üriner anomalilerin, hidrosefalinin, kongenital kardiak anomalilerin ve exteremite malformasyonlarının birlikte görülebileceği genetik bir bozukluktur.

- Teli M, Hosalkar H, Gill I, Noordeen H. Spondylocostal dysostosis: thirteen new cases treated by conservative and surgical means. Spine (Phila Pa 1976). 2004;29:1447-51.
- Berdon WE, Lampl BS, Cornier AS, Ramirez N, Turnpenny PD, Vitale MG, Seimon LP, Cowles RA. Clinical and radiological distinction between spondylothoracic dysostosis (Lavy-Moseley syndrome) and spondylocostal dysostosis (Jarcho-Levin syndrome). Pediatr Radiol. 2011;41:384-8.
- Solomon L, Jimenez RB, Reiner L. Spondylothoracic dysostosis: report of twocases and review of the literature. Arch Pathol Lab Med. 1978;102:201-5.

JARCHO-LEVİN SENDROMU

- İlk olarak 1938’de Jarcho ve Levin tarafından porto-riko’lu bir ailede tanımlanmıştır.

- Otozomal dominant ve otozomal resesif kalıtımla bildirilmiştir.

- Hem akraba evliliklerinde hem de akraba olmayan ailelerde bildirilmiştir.

- Erkekler ve kızlar eşit sıklıkta etkilenmektedir

Jarcho S, Levin PM. Hereditary malformations of the vertebral body. Bull John Hopkins Hosp. 1938;62:216–226.

Kulkarni ML, Sarfaraz Navaz R. Jarcho-Levin Syndrome – Clinical Brief. Indian Journal of Pediatrics. 2006;73:73–75.

JARCHO-LEVİN SENDROMU

- Görülme sıklığı 25/10000 oranındadır.
- Prognozu respiratuvar problemler ile ilişkili olarak değişmektedir.

- Spondilokostal (SCD)
- Spondilotorasik (STD)

→ olarak ikiye ayrılır

- Berdon WE, Lampl BS, Cornier AS, Ramirez N, Turnpenny PD, Vitale MG, Seimon LP, Cowles RA. Clinical and radiological distinction between spondylothoracic dysostosis (Lavy-Moseley syndrome) and spondylocostal dysostosis (Jarcho-Levin syndrome). Pediatr Radiol. 2011;41:384-8.

- Solomon L, Jimenez RB, Reiner L. Spondylothoracic dysostosis: report of twocases and review of the literature. Arch Pathol Lab Med. 1978;102:201-5.

JARCHO-LEVİN SENDROMU

- SCD hem otozomal dominant hem de resesif kalıtımla kalıtılabilir. Hastaların DLL3, MESP2 ve LFNG genlerinde mutasyona sahip olduğu bilinmektedir.

- STD alt tipi, otozomal resesif geçişli bir bozukluk ve MESP2 geninde mutasyonlar bulunur.

- Kulkarni ML, Sarfaraz Navaz R, Vani HN. Jarcho-Levin Syndrome – Clinical Brief. Indian Journal of Pediatrics. March 2006; 73:73-75

- Whittock NV, Sparrow DB, Wouters MA, Sillence D, Ellard S, Dunwoodie SL, Turnpenny PD. Mutated MESP2 Causes Spondylocostal Dysostosis in Humans. Am J Hum Genet. June 2004; 74(6): 1249–1254

Materyal ve Metot

- Mayıs 2012 ile Kasım 2014 tarihleri arasında yatırılan ve JLS saptanan olgular alındı.

- Tüm olguların detaylı fizik muayeneleri yapılarak yenidoğan ünitesine yatırıldı.

- Eşlik eden anormal bulgular kaydedildi.

- Transfontanel USG'de anormallik saptanan tüm olgulara ileri kranial görüntüleme yapıldı.

Sonuçlar

- Çalışmaya 12 olgu alındı.
- Olguların tamamı doğumda ciddi solunum sıkıntısı nedeniyle solunum desteği aldı.
- Mortalite sıklığı % 41.7 (4 ST, 1 olgu SC)
- Arnold Chiari malformasyonu tip 2 —> 12 olgu
- Corpus callozum disgenezisi —> 6 olgu
- MMS —> 12 olgu
    - Lumbosakral—> 6 olgu,
    - Lomber —> 3 olgu
    - Torakal —> 3 olgu
- Holoprozensefali —> 1 Olgu
- Porensefalik kist —> 1 olgu

Hastalarımızın demografik özellikleri

Table 1. Key Clinical Data.

Patient no.	Sex	Gestation age (week)	Length (cm)	Weight (g)	Head circum (cm)	Primary diagnosis	Relationship	Prognosis	Anomaly				
									Costal/vertebral	CNS	CVS	GUS/anorectal	Skeleton
1	Female	40	52	3500	50	JLS	None	Exitus	+/+	LS-MMS + ACM2	ASD	ND	S
2	Female	39	48	3000	40	JLS	First cousin	Live	+/+	LS-MMS + ACM2 + HPE	PFO	ND	S + K
3	Male	39	49	3000	39	JLS	None	Exitus	+/+	LS-MMS + ACM2	ND	ND	S + K + PE
4	Female	40	44	3050	42.5	JLS	Third degree	Live	+/+	T-MMS + ACM2 + CCD	PDA + ASD	ND	S + K + PE
5	Male	39	40	2100	38	JLS	First cousin	Live	+/+	T-MMS + ACM2	ASD	Unilateral kidney	S + K
6	Male	39	43	2720	37	JLS	None	Live	+/+	LS-MMS + ACM2 + CCD	PFO	ND	ND
7	Female	39	42	2900	35	JLS	First cousin	Live	+/+	LS-MMS + ACM2 + CCD	ASD + PDA	ND	S + PE
8	Female	39	42	2420	35	JLS	First cousin	Exitus	+/−	L-MMS + PC	Dextrocardia + VSD + ASD	AG	AMK
9	Male	39	39	3270	35.5	JLS	First cousin	Live	+/+	L-MMS + ACM2 + CCD	PFO	AA	S + K + PE
10	Female	39	46	4300	38	JLS	First cousin	Exitus	+/+	T-MMS + ACM2	ND	AA	ND
11	Female	39	42	2500	38	JLS	None	Live	+/+	LS-MMS + ACM2 + CCD	ASD	ND	S + K
12	Male	39	49	3200	45	JLS	None	Exitus	+/+	L-MMS + ACM2 + CCD	PFO	ND	S + K

AA, anal atresia; ACM2, Arnold–Chiari type 2 malformation; AG, ambiguous genitalia; AMK, arthrogryposis multiplex konjenita; ASD, atrial septal defect; CCD, corpus callosum dysgenesis; CNS, central nervous system; CVS, cardiovascular system; GUS, genitourinary system; HPE, holoprosencephaly; JLS, Jarcho–Levin syndrome; K, kyphosis; L-MMS, lumbal meningomyelocele; LS-MMS, lumbosacral meningomyelocele; ND, not defined; PC, porencephalic cyst; PDA, patent ductus arteriosus; PE, pes equinovarus; PFO, patent foramen ovale; S, scoliosis; T-MSS, thoracic meningomyelocele; VSD, ventricular septal defect; +, present; −, absent.

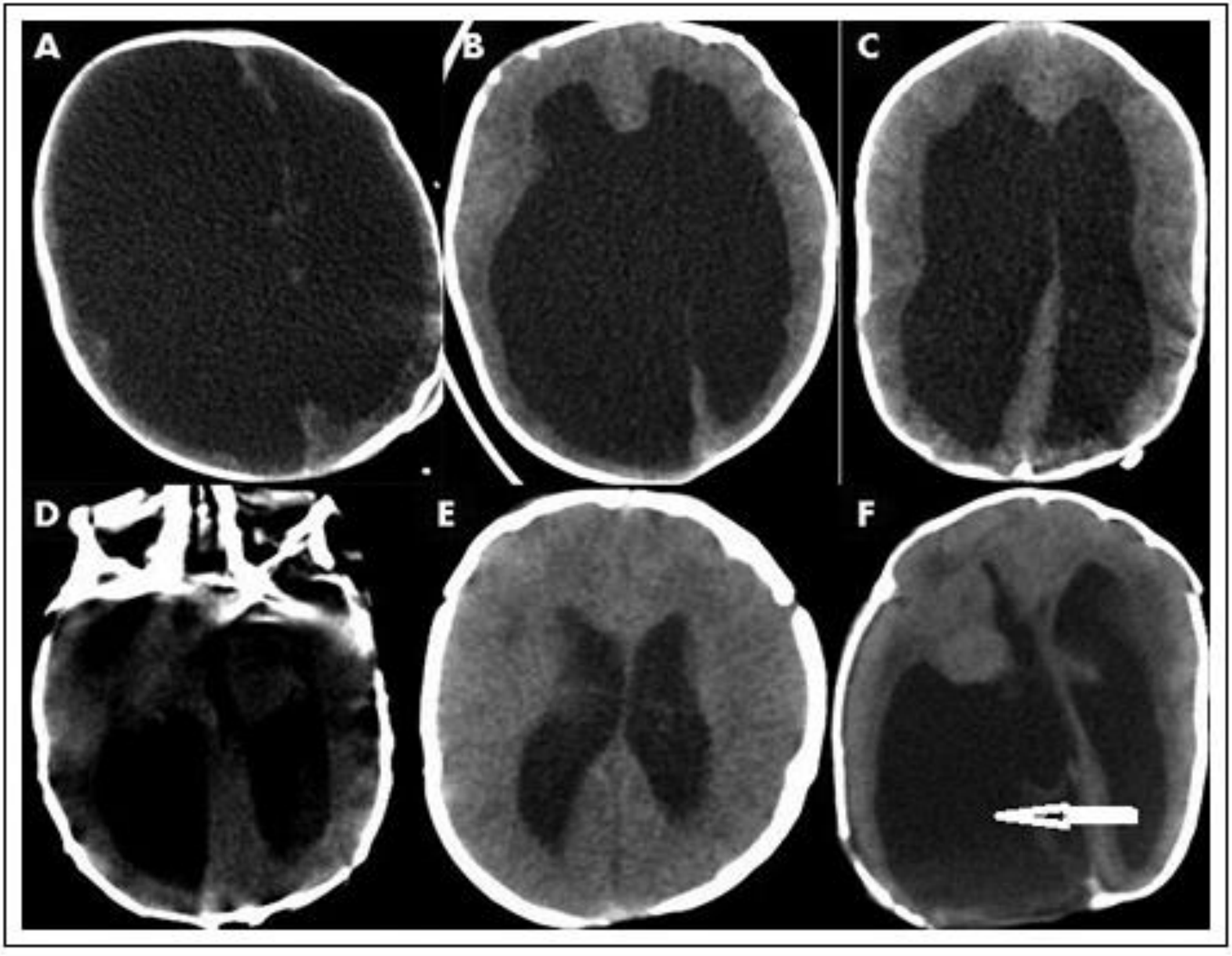
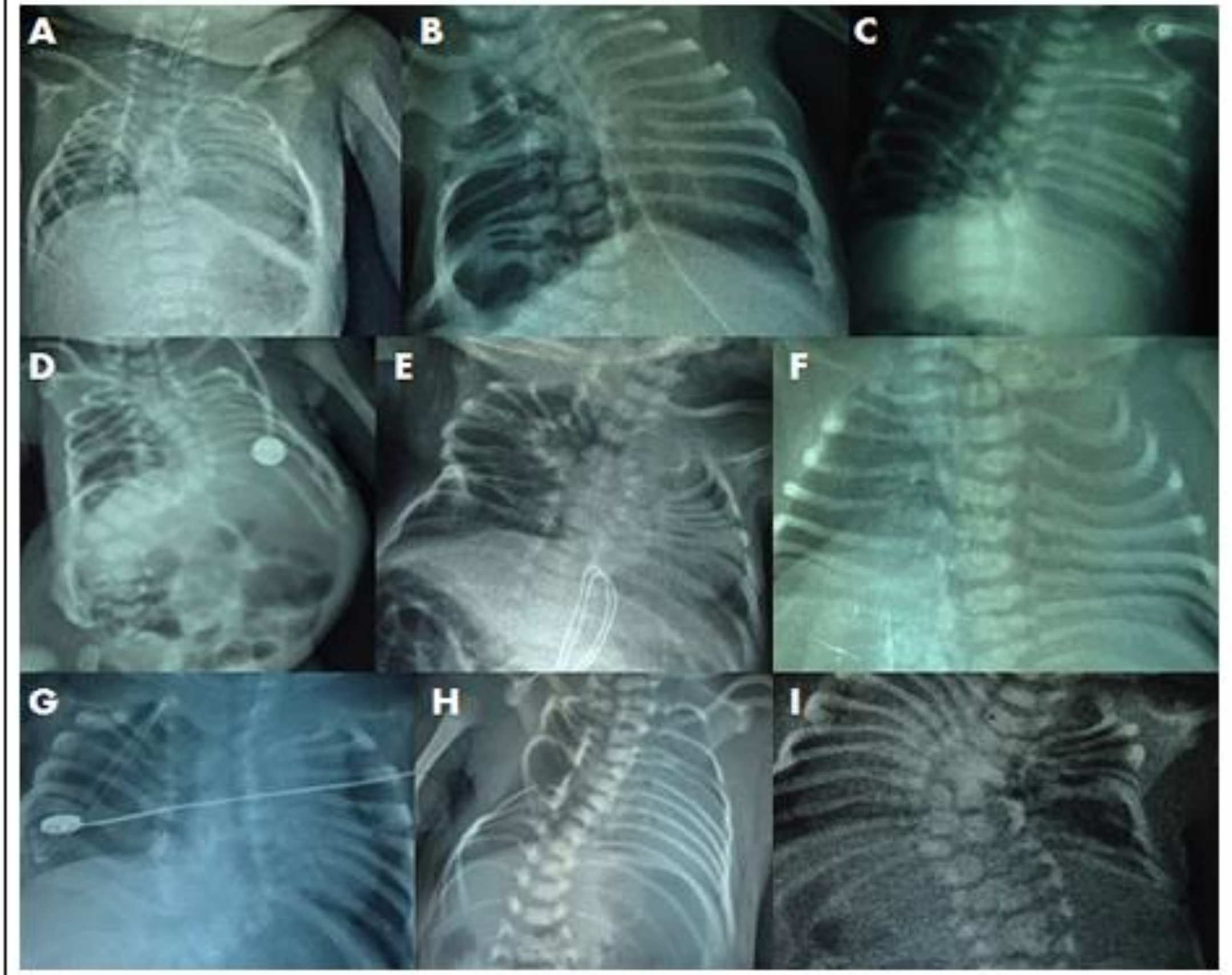


Figure 1. Axial computed tomography images of the patients revealed massive hydrocephalus (A, C, D, E); holoprosencephaly (in patient 2) (B); porencephalic cyst (44 × 80 mm) (in patient 8) (F).


A
B
C
D
E
F
G
H
I

Amaç

Bu çalışmada JLS'li olgulara Santral Sinir Sistemi anomalilerinin sıklıkla eşlik ettiğini vurgulamaktır

Tartışma

- NTD'li hastaları kapsamlı bir şekilde inceleyerek ilişkili anomalilerin erken tanı ve tedavisi hayati önem taşımaktadır.

- JLS'de göğüs deformitesinden dolayı akciğer fonksiyonları bozulmaktadır.

- Bu durum enfeksiyonların daha kolay gelişmesine yol açmaktadır.

- Bu yüzden tedavide agresif davranılmalıdır.

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JARCHO-LEVIN SYNDROME PRESENTING WITH SEVERE HYDROCEPHALUS: A REPORT OF THREE CASES

BY S. YURTTUTAN [1], H. DEĞIRMENCIOĞLU [1], M.Y. ONCEL [1], S. KARA [2], F. SARI [1] AND U. DILMEN [1,3]

Summary: *Jarcho-Levin syndrome presenting with severe hydrocephalus: a report of three cases:* Jarcho-Levin syndrome (JLS) is a genetic disorder characterized by defects of spine formation, abnormal fusion of the ribs at the costovertebral and costochondral junction due to abnormal vertebral and costal segmentation. There are two subtypes associated with different distribution and severity of defects in JLS. While vertebral segmentation abnormalities are frequent in spondylo-thoracic subtype costal segmentation and fusion abnormalities are frequent in spondylo-costal subtype. Neural tube defects with severe hydrocephalus are rare findings in this syndrome. Herein we report three infants with severe hydrocephalus associated with Jarcho-Levin syndrome.

Key-words: Jarcho-Levin syndrome – Hydrocephalus – Newborn.

3 vakalık bu çalışmada Hidrosefali ve NTD'nin bu olgularda nadir olduğu vurgulanmıştır.

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Perinatal Diagnostic Approach to Fetal Skeletal Dysplasias: Six Years Experience of a Tertiary Center

Havva Serap Toru, Banu Guzel Nur, Cem Yasar Sanhal, Ercan Mihci, İnanç Mendilcioğlu, Elanur Yilmaz, Gulden Tasova Yilmaz, Irem Hicran Ozbudak, Kamil Karaali, Ozgul M. Alper & Fatma Şeyda Karaveli

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To link to this article: http://dx.doi.org/10.3109/15513815.2015.1068414

Toru ve ark. Perinatal ve fetal otopsi yapılan 542 vakanın 54'ünde iskelet displazisi saptamış ve bu hastalarda JLS insidansını % 1.9 olarak saptanmıştır.

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Table 1. Individuals with SCD and accompanying central nervous system malformations

Patient	MMC	HC	SC	CM	TC	SYRM	MCM
1			+		+	+	+
2	+	+					
3	+		+				
4	+					+	
5	+	+				+	
6			+				
7	+	+					
8	+						
9	+						
10	+	+		+			
11	+		+				
12	+						
13	+	+					
14	+	+		+			
15	+						
16	+	+					
17	+		+	+			
18	+						
19	+		+			+	
20	+	+					
21	+	+					
22	+	+					
23	+	+					
24	+	+	+	+	+		
25	+	+					
26	+	+					
27	+	+					
28	+	+					

HC = Hydrocephalus; SC = split cord; CM = Chiari malformation; TC = tethered cord; SYRM = syringomyelia; MCM = mega cisterna magna.

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A Single-Center Experience of CNS Anomalies or Neural Tube Defects in Patients With Jarcho–Levin Syndrome

Nihat Demir, MD[1], Erdal Peker, MD[1], İsmail Gülşen, MD[2], Kemal Ağengin, MD[3], Sultan Kaba, MD[4], and Oğuz Tuncer, MD[1]

Abstract

Jarcho–Levin syndrome (JLS) is a genetic disorder characterized by distinct malformations of the ribs and vertebrae, and/or other associated abnormalities such as neural tube defect, Arnold–Chiari malformation, renal and urinary abnormalities, hydrocephalus, congenital cardiac abnormalities, and extremity malformations. The study included 12 cases at 37-42 weeks of gestation and diagnosed to have had Jarcho–Levin syndrome, Arnold–Chiari malformation, and meningmyelocele. All cases of Jarcho–Levin syndrome had Arnold–Chiari type 2 malformation; there was corpus callosum dysgenesis in 6, lumbosacral meningmyelocele in 6, lumbal meningmyelocele in 3, thoracal meningmyelocele in 3, and holoprosencephaly in 1 of the cases. With this article, the authors underline the neurologic abnormalities accompanying Jarcho–Levin syndrome and that each of these abnormalities is a component of Jarcho–Levin syndrome.

Keywords

Jarcho–Levin syndrome, neural tube defect, Arnold–Chiari type 2, newborn

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Sonuç olarak

- JLS ile NTD birlikteliği oldukça sıktır

- JLS'nin Spondilotorakal Dizostozis alt grubunda ciddi solunum problemleri olur.

- Prenatal tanıda hastalar değerlendirilirken JLS düşünülen olgularda SSS anomalileri ve kostovertebral malformasyonların irdelenmesi gerekir

- Hazırlık için YD ekibinin bu konuda bilgilendirilmesi önemlidir.

Teşekkürler