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Annals of Pediatrics & Child Health

Research Article

Cephalopolysyndactyly Orofacial Clefts Disorders of Sexual Development Renal Anomaly with Hirschsprung's Disease an unexpected Spectrum of Anomalies under Chromosome Partial Monosomy 7 with a deletion in the p21.3 segment: Clinical Image

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Submitted: 10 February 2023

Accepted: 14 March 2023

Published: 16 March 2023

ISSN: 2373-9312

Keywords

- Monosomy 7
- Deletion
- p21.3 segment
- Anomalies

Abstract

Chromosome microdeletion is a hereditary disease with complex clinical manifestations due to the deletion of small chromosome segments [1]. Chromosome 7, Partial Monosomy 7p is a rare chromosomal disorder characterized by deletion (monosomy) of a portion of the short arm (p) of chromosome 7 (7p) [2]. Associated symptoms and findings may be variable and may depend on the specific size and location of the deleted segment of 7p [2].

KEY CLINICAL MESSAGE

In this image report, we highlight a case of abnormal karyotype and chromosome 7 with a deletion in the p21.3 segment carrying an expected and rare spectrum of pathologies were they pose a diagnostic problem to the clinician.

CLINICAL IMAGE

A 4 years -old male has been followed by our pediatric surgery clinic since birth as a high-risk patient follow-up. He had been diagnosed with intrauterine growth restriction (normal vaginal delivery, birth weight 2.170 kg, height 41 cm) at 37 weeks gestation. A product for G5 P0 A0 mother, no familial disease or history of maternal infection. In the process of follow-up physical examination, we found that his weight, height, and head circumference were all below the 3rd percentile. Multidisciplinary clinician teams arranged the patient for a dysmorphology examination to pick up the anomalies that are readily apparent. Abnormal shape of the head, and lack of cranial vault growth, upper and lower limbs polydactyly (Figure 1, A-E). Small phallus with an underdeveloped scrotum, with evidence of recurrent urinary tract infection (Figure 2, A-G). Cephalopolysyndactyly manifestation pointed the patient for karyotype and chromosomal studies (Figure 3, A-E). Delayed passage of meconium and chronic constipation with abdominal distention arranged him for further assessments (Figure 4, A-D). Our patient followed precisely under a protocol of management.

DISCUSSION

Chromosome 7 contains more than 158 million base pairs and represents approximately 5% of total cellular DNA [3]. Partial Monosomy 7p may vary in range and severity from case to case; many affected individuals have growth delays before and after birth (prenatal and postnatal growth retardation) [2,3]. Other physical and pathological features, such as growth deficiency, musculoskeletal abnormalities, genital defects, structural malformations of the heart that are present at birth (congenital heart defects), and/or another spectrum of abnormalities may associate with monosomy syndrome. In many cases, there is the early closure of the fibrous joints (cranial sutures) between certain bones of the skull (craniosynostosis), additional reported findings have included a highly arched roof of the mouth (palate)

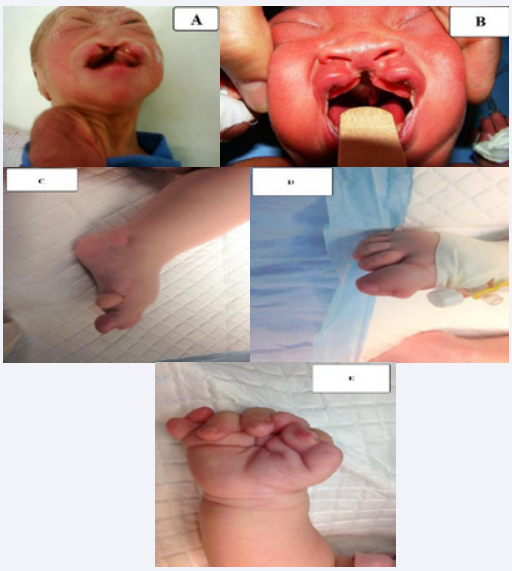

Figure 1 A-E: A&B; Abnormal shape of the head, lack of cranial vault growth, down slanting eyelid folds (palpebral fissures), syndromic facial dysmorphisim , asymmetric crying face, and orofacial clefts with bifid uvula. C&D; Preaxial (extra hallux) and postaxial polydactyly (extra fifth toe) of the right and left feet. E; Preaxial polydactlyly (thumb duplication) and postaxial polydactlyly (small finger duplication) of the right hand (polysyndactyly).

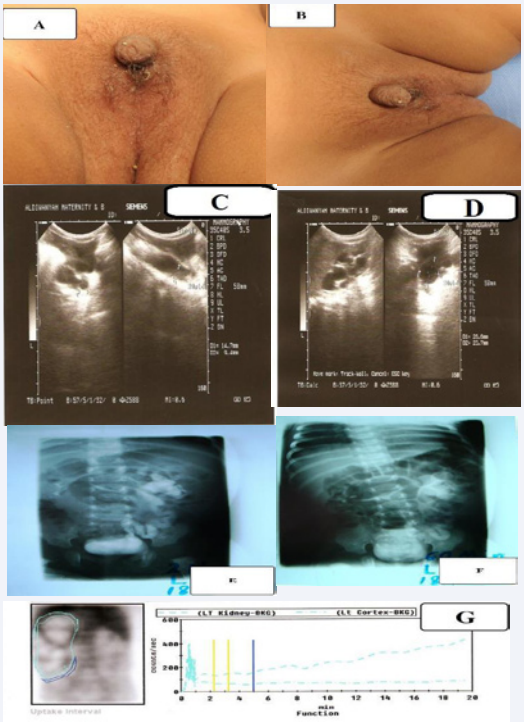

Figure 2 A-G: A&B; Small phallus with bilateral undescended testis and underdeveloped scrotum (disorders of sexual development). C&D; Abdominal ultrasound study revealed a submucosal cystic dilation of the terminal segment of the left ureter (ureterocele). E&F; An intravenous urogram (IVU) reported a round radiopacity in the bladder surrounded by a radiolucent rim (intravesical ureterocele of left ureter) with dilatation of the ureter and renal pelvis. G; Diuretics dynamic renal scan with Tc-99m DTPA revealed the left kidney with poor parenchymal uptake and excretion (split function 25%).

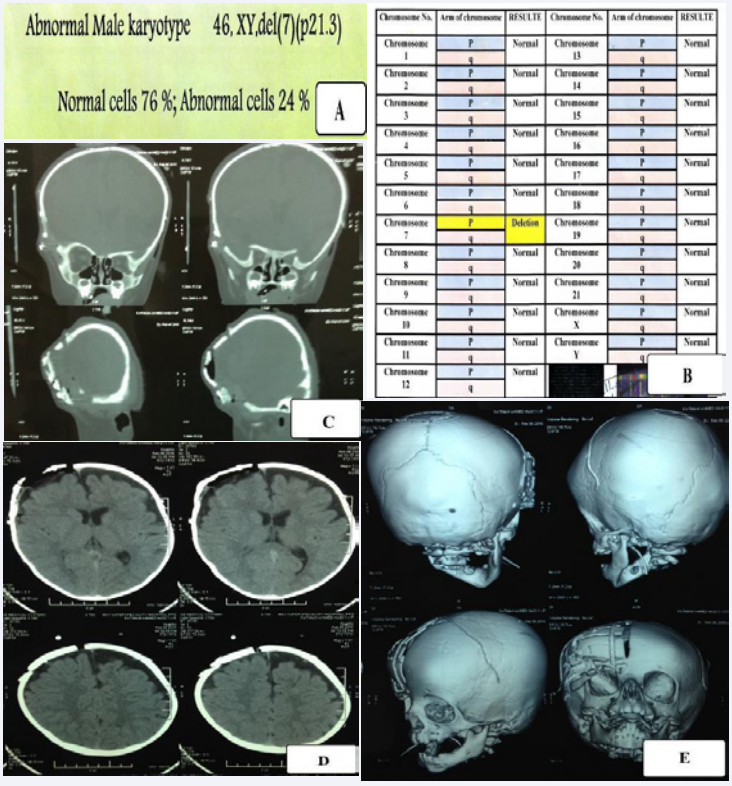

Figure 3 A-E A&B; karyotype and chromosomal study by interphase fluorescence in situ hybridization submitted abnormal male karyotype with distal deletions to the terminal band of chromosome 7, p21.3 with abnormal cell. C&D; Preoperative computerized tomography scan (CT) of the patient skull demonstrating closed sagittal suture (scaphocephaly). E; Post-operative 3-dimensional (3D) CT scans of the skull revealed reshaped the affected portion of the skull and the skull position was held in place with plates and screws.

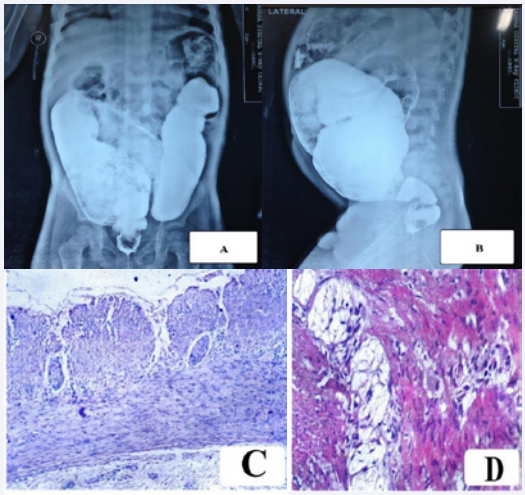

Figure 4 A-D: A; Anteroposterior barium enema study of the same patient revealed showing a markedly dilated, feces-filled colon. B; Lateral film revealed reduced caliber of the rectum, a transition zone with an enlarged-caliber sigmoid. C&D; Hematoxylin-eosin (HE) staining of a full-thickness rectal wall fragment histopathology studies submitted a lack of ganglion cells in the submucosal or intramuscular nerve plexus of the intestinal wall and the presence of hypertrophic nerve fibers and trunks.

or incomplete closure (cleft palate) [2]. Reported features have included permanent flexion of one or more fingers, unusually short hands; abnormalities of the thumbs. Underdevelopment (hypoplasia) of the external genitals, urological anomalies with an abnormally small colon (micro colon), 7p deletion is a recurrent cytogenetic abnormality and or other defects were reported [3&4]. Works of literature submitted, there is significant variation in the size and location of the deleted segment of 7p, accordingly may affect the range and severity of associated symptoms and findings [4].

CONCLUSION

For the clinician, chromosome partial monosomy 7 with a deletion in the p21.3 segment with such spectrum carry a challenge for the management protocol and may minimize the horizon of the outcome.

CONFLICT OF INTEREST

The authors declare that they have no competing interests. This work is original and not submitted with other publishers.

ETHICS STATEMENT

informed consents were obtained from the patients› parent who participated and managed in this report for publication and any accompanying images. The report conformed to the guidelines of the institutional review board of our Institution, which approved its ethical aspects.

ACKNOWLEDGMENTS

The author expresses sincere gratitude to all the pediatric surgery unit staff, pediatric outpatient's clinic, and the department of radiology at the Maternity and Child Teaching Hospital, Al Qadisiya- Al Diwaniya, Iraq, for their assistance. We are grateful to all colleagues in Al Bailasan lab, for their kind assistance to conclude the karyotype, chromosomal and the histopathology results and images.

AUTHOR CONTRIBUTION STATEMENT

MA conceived and designed the study. MA and SK were responsible for data collection and acquisition of data; they wrote the initial and revised the report. All authors have approved the final manuscript.

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