Case Report

Prune Belly Syndrome Associated with Interstitial 17q12 Microdeletion

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We report a term male neonate presenting with a “prune belly,” bilateral hydronephrosis, hydroureter, posterior urethral obstruction, and bilateral undescended testes. Analysis with the whole genome SNP microarray revealed an interstitial deletion of about 1.49 megabase (MB) at chromosome 17q12. We present a rare association of prune belly syndrome with a chromosomal deletion in this same region.

1. Introduction

Interstitial deletions involving terminal 17q12 (17q12-qterm) are rare chromosomal abnormalities associated with variable phenotypes. Previous case reports of 17q12 microdeletion characterized the patients with structural and/or functional disorders of the kidney and urinary tract, maturity-onset diabetes of the young type 5 (MODY5), and neurodevelopmental or neuropsychiatric disorders (e.g., developmental delay, intellectual disability, autism spectrum disorder, schizophrenia, anxiety, and bipolar disorder) [1–3]. To date, there are few reported cases of prune belly syndrome (PBS) and associated chromosomal abnormalities. We report a new case of PBS associated with 17q12 microdeletion, and we review the literature.

2. Case Report

A term Hispanic male neonate was born at 40-week gestation to an 18-year-old primigravida by cesarean section. Apgar scores were 9 and 9 at 1 and 5 minutes, respectively. Pregnancy was complicated by an abnormal prenatal ultrasound at 20-week gestation which revealed bilateral renal pelvis dilation, dilated ureters, thickened bladder with key-hole sign, and dilated urethra. Subsequent sonography revealed progression of the hydronephrosis and hydroureters. Genetic studies that were performed during the pregnancy had shown 17q12 microdeletion, a de novo mutation. The family history was negative for congenital anomalies, and there were no history of in utero exposure to any known teratogens and no history of consanguinity.

Physical examination revealed a weight of 3470 grams (45th centile), length of 50 cm (40th centile), and head circumference of 35 cm (45th centile). Anomalies noted at birth included marked abdominal distension with a thin, wrinkled, and flaccid abdominal wall and bilateral undescended testes (shown in Figures 1 and 2). A Foley catheter was placed on day of life (DOL) 0, and the infant was started on amoxicillin for urinary tract infection (UTI) prophylaxis. Ultrasound of the kidneys and bladder on DOL 0 showed severe right hydroureteronephrosis, moderate left hydroureteronephrosis, thick-walled urinary bladder, and dilatation of the posterior urethra. There was a cystic structure noted in the prostate (shown in Figure 3). An abdominal ultrasound found bilateral testes in the midabdomen. Voiding cystourethrogram (VCUG) performed on DOL 2 revealed grade 5 left vesicoureteral reflux and grade 1 right vesicoureteral reflux. The cystic structure in the prostate represented the prostatic utricle cyst on VCUG. The posterior urethral
dL. The infant's feeds consisted of breastmilk or Similac and orchiopexy at 1 month. Upon discharge on DOL 21, serum creatinine was 0.8 mg/dL and orchiopexy at 1 month. After the procedures, he developed pyelonephritis despite UTI prophylaxis.

3. Cytogenetic and Molecular Studies

Whole genome SNP (single-nucleotide polymorphism) microarray analysis was performed using the SNP oligonucleotide microarray analysis (SOMA) CytoScan HD platform which uses over 743,000 SCN probes and 1,953,000 NPCN probes with median spacing of 0.88 kilobase (kb). Total genomic DNA was extracted from the patient's blood sample and digested with NspI and ligated to NspI adaptors. Polymerase chain reaction (PCR) products were purified and quantified. Purified DNA was fragmented and biotin labeled and hybridized to the CytoScan HD GeneChip. There was a 1.49 megabase (MB) interstitial deletion of the long arm of chromosome 17: arr [hg19] 17q22 (34,822, 465-36, 307,773) x1 and 826 kilobase (kb) interstitial duplication in the long arm of chromosome 7: 7q12.1 (98,230,688-99, 056,822) x3.

The SNP microarray analysis identified an interstitial deletion of 17q22.1, and this deleted interval includes numerous Online Mendelian Inheritance in Man (OMIM) genes starting from ZNHT3 to TBC1D3H. Deletion of this region has been reported to be associated with the following phenotypes: cystic renal disorders, pancreatic atrophy, liver abnormalities, cognitive impairment and structural brain abnormalities, maturity-onset diabetes of the young (MODY), Müllerian aplasia/Mayer-Rokitansky-Küster-Hauser syndrome in females, and epilepsy (OMIM 189907).

Whole genome SNP microarray (Reveal) analysis identified an interstitial duplication of 7q12.1; this interval includes 10 OMIM genes (NPTX2, TRRAP, SMURF1, KPNA7, ARPC1A, ARCP1B, PDA1, BUD31, PTCD1, and CPSF4). There has been no report of clinically established disorders with duplication of this region.

4. Discussion

Aplasia of the abdominal wall muscle was described by Froehlich in 1939, which is considered the first description of PBS [1]. The incidence of PBS is estimated about 1/30,000 to 1/50,000 live births [2–4], with the incidence decreasing likely due to improvement of prenatal detection and elective termination of affected pregnancies. Most of the cases of PBS are male (3-5% being female), prevalence is higher in the black population, and incidence is higher in infants born to younger mothers [5, 6]. The triad of PBS manifestation included (1) dygenesis and partial or complete aplasia of muscle of the abdominal wall, (2) undescended testes, and (3) complex malformations of the urinary tract [7].

There are other findings apart from the triad, which are present in 75% of the PBS cases. Pulmonary abnormalities, e.g., pulmonary hypoplasia (not parenchymal lung disease), are present and related to prenatal oligohydramnios. Gastrointestinal complications included malrotation, intestinal atresia/stenosis, volvulus, and obstruction occurring in about 30% of PBS patients [8]. 10% of PBS cases have associated cardiac anomalies: septal defects, tetralogy of Fallot, and patent ductus arteriosus [3]. Musculoskeletal abnormalities, usually lower extremities: club feet, congenital hip dislocation, and hypoplasia of the leg or foot, were seen in 5% of the PBS cases [2, 9].

The pathogenesis of PBS has been debated and currently proposed with two prominent thoughts: mesenchymal developmental failure or urinary tract obstruction [10]. Higher incidence in males and cases of familial PBS lead to the speculation of a possible sex-influenced inheritance pattern. Murray et al. and Haeri et al. reported 2 cases of PBS
with interstitial deletions of chromosome 17q12, suggesting the role of \textit{HNF1B} (\textit{TCF2}), a gene that is responsible for mesodermal and endodermal development in different tissues, in PBS [11–13]. However, 17q12 deletion has also been reported in association with other congenital abnormalities of the kidney and urinary tract (CAKUT) without the PBS phenotype: agenesis, hypoplasia, dysplasia, multicystic dysplastic kidney (MCDK), and horseshoe kidney, collecting system abnormalities (duplicated collecting systems, ureteropelvic junction obstruction, isolated hydronephrosis, or hydroureter) and tubulointerstitial disease of the kidney [14–18].

\textbf{5. Conclusion}

We report a case of a neonate with 17q12 microdeletion that is associated with PBS. Our report supports the genetic basis in PBS, and the screening for \textit{HNF1B} gene mutations/deletions on chromosome 17q12 could help identify the patient.
of PBS and lead to preventing and improving the treatment of this rare disease.

**Data Availability**

All data pertaining to this article are available from the authors.

**Consent**

Written informed consent was obtained from the parents of the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the editor-in-chief of this journal.

**Conflicts of Interest**

The authors declare no conflict of interest.

**Authors’ Contributions**

All authors are equally involved in drafting, literature search, and writing the paper.

**References**


