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CASE REPORT

Road to a rare diagnosis: Description of novel unbalanced translocation causing partial trisomy 17p

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Abstract

Trisomy 17 is a rare chromosomal disorder. Existing literature on the topic is limited and mostly refer to mosaic Trisomy 17 cases. Our report summarizes the 70-day clinical course of a late preterm neonate with partial Trisomy 17p karyotype 46,XY,der(14)t(14;17)(p11.1;p11.2) dpat. Trisomy 17 due to unbalanced translocation is rare, and our case elaborates the clinical presentation with intestinal malfunction without any anatomical pathology and urethral diverticulum and the ethical dilemma in decision-making. The male proband was born at 35 weeks with antenatal findings of multiple neurological and other abnormalities such as cystic hygroma, absent corpus collosum, high riding third ventricle, absent cavum septum pellucidum, indented occiput, absent ductus venous, and intrauterine growth restriction. The postnatal findings included significant facial dysmorphisms with short palpebral fissures, hypertelorism, low set ears, micrognathia, hirsutism, and single palmar creases, central hypotonia, and hyperreflexia of upper limbs bilaterally. Genital-urinary assessment revealed a urinary diverticulum and significantly underdeveloped scrotum with undescended testes. Infant had excessive irritability and resistance to sleep despite increasing doses of analgesia and sedation, and persistent respiratory and feeding difficulties. Enteral nutrition could not be established due to profuse and persistent diarrhea, necessitating use of total parenteral nutrition. Microarray assay exhibited a pathogenic copy number gain of approximately 21.4 Mb of chromosome region 17p13.3p11.2. Follow-up chromosome analysis and FISH revealed an abnormal male karyotype with a derivative chromosome 14, resulting from an unbalanced translocation between the short arm of one chromosome 14 and the short arm of one chromosome 17, effectively resulting in trisomy 17p11.2. It was derived from a paternal balanced t(14;17)(p11.1;p11.2) as shown by chromosome analysis and FISH studies. The rarity of this chromosomal disorder contributed to difficulty with prognosis and led to bioethical dilemma regarding life-sustaining measures and quality of life. Through shared decision-making processes and in consideration of poor

prognosis, parents decided to withdraw life-sustaining care and the proband died at postnatal day of life 70.

KEYWORDS

derivative chromosome 14, partial trisomy 17p, unbalanced translocation

1 | INTRODUCTION

Trisomy 17 is a rare genetic disorder. There is limited information available about antenatal findings, phenotype, clinical management options, and outcome in live-born infants.

In the reported cases neurological malformations (e.g., microcephaly, ventriculomegaly, cerebellar hypoplasia, and hypoplastic cerebellar vermis), congenital heart defects,[1] pre- and postnatal growth retardation,[2] clinodactyly of fingers, and psychomotor and mental delays[3,4] are described. Prognosis remains guarded for this disorder. We report unique urethral and genital anomalies in context of a patient with trisomy 17p due to unbalanced translocation involving chromosomes 14 and 17. The translocation was paternal in origin.

2 | CLINICAL REPORT

The proband was a male newborn, of natural conception born at 35 2/7 weeks, to a 34-year-old (GTPAL:21001) mother, and a 42-year-old father. Both parents are of Middle Eastern descent, and reportedly non-consanguineous. There is a 6-year-old healthy sister. Family history was significant for two neonatal demises in the first 2 months of life from the maternal grandmother side and two miscarriages from the paternal grandmother side. This pregnancy was remarkable for cystic hygroma seen in the first trimester with persistent intra-uterine growth restriction (IUGR) throughout the pregnancy. Multiple detailed anatomy scans revealed an absent corpus callosum, high riding third ventricle, absent cavum septum pellucidum, and indented occiput.

Fetal echocardiography showed an absent ductus venosus and drainage directly to the hepatic vasculature without other intracardiac anomalies. The non-invasive prenatal testing (NIPT) was low risk for or trisomy 13, 18, and 21 and monosomy X. The couple was counseled and offered the option of amniocentesis; however, they declined. The proband was born via caesarean section due to sustained fetal heart decelerations during non-stress testing. Apgar scores were 9 and 9 at 5 and 10 min respectively. Nasal continuous positive airway pressure (NCPAP) was initiated due to persistently low oxygen saturation and transferred to the NICU. He was small for gestational age on all parameters: birth weight was 1510 g (1st percentile), length was 38.5 cm (1st percentile), and occipitofrontal circumference (OFC) was 30 cm (8th percentile).

Physical examination was significant for a small for gestational age baby, short palpebral fissures, hypertelorism, low set ears, micrognathia, hirsutism, flat nasal bridge, elongated philtrum, thin upper lip, anteverted nares (Figure 1A), and a single palmar crease in both hands. His neurological examination was consistent with central hypotonia, bilateral hyperreflexia of upper limbs, and a sacral dimple with a tuft of hair.

On genital-urinary examination, there was an underdeveloped scrotum with bilateral palpable undescended testicles. The external genitalia examination showed penile phallus with an incomplete foreskin. The distal aspect of the penile shaft was enlarged proportional to the proximal aspect. A pinpoint hypospadic (coronal) meatus was visualized (Figure 1B– red circle), however was not catheterizable despite multiple attempts.

The proband remained in NICU for 70 days and experienced multiple significant medical issues. Neurologically, he had sleeping difficulty and irritability, which became pronounced with advancing postnatal age, mandating the need for multiple analgesics and sedatives, including acetaminophen, chloral hydrate, morphine, and benzodiazepines. Brain magnetic resonance imaging (MRI) revealed absent corpus callosum and septum pellucidum cavum with associated parallel colpocephalic lateral ventricles and a small optic chiasma. Sacral spine ultrasound was negative. Electroencephalogram at 4 weeks revealed poorly differentiated low voltage background without any epileptiform activity. There were positive phase reversal sharp transients over the temporal regions, which could have suggested a deep white matter injury or a diffuse cerebral dysfunction.

The proband also demonstrated unexpected and prolonged dependence on respiratory support (NCPAP and high-flow nasal canula oxygen) his entire life suspected to be central in nature given low supplemental oxygen requirements and lack of significant radiological parenchymal findings.

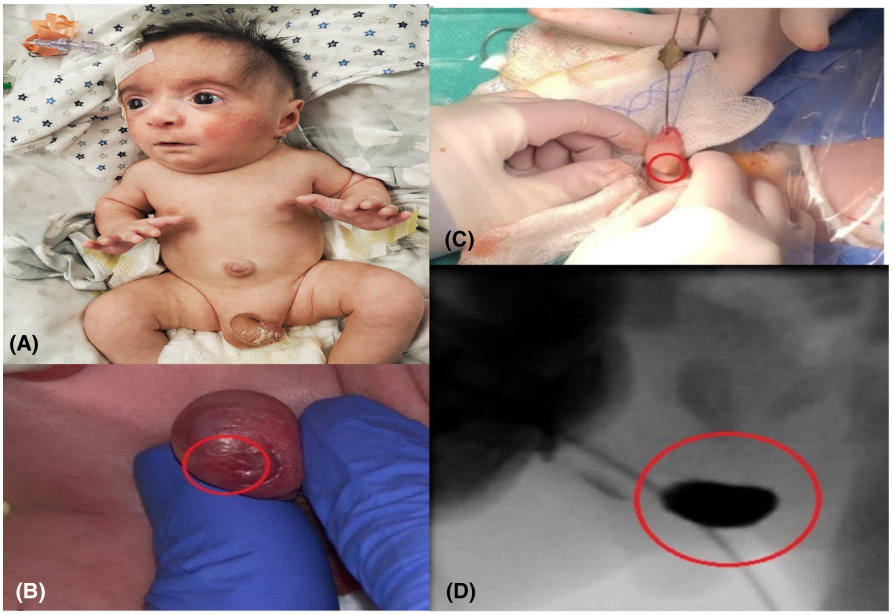

FIGURE 1 (A) short palpebral fissure, hypertelorism, low set ears, micrognathia, flat nasal bridge, elongated philtrum, thin upper lip, anteverted nares (B) A pinpoint hypospadic (coronal) meatus was visualized (red circle) (C) The urethral diverticulum confirmed using a lacrimal probe (red circle) (D) distal penile swelling indicated urine pooling within a urethral diverticulum on contrast study (red circle)

From the cardiovascular perspective, initial echocardiogram after birth showed a small apical ventricular septal defect (VSD), moderate size patent ductus arteriosus (PDA), small secundum atrial septal defect (ASD), and left superior vena cava (SVC) to coronary sinus with signs of elevated pulmonary arterial pressure. Subsequent echocardiogram showed closure of PDA without any evidence of progressive cardiomegaly or cardiac dysfunction.

Gastrointestinal (GI) issues were significant for persistent and severe feeding intolerance presenting as emesis and feed-associated irritability despite the absence of any identifiable anatomical abnormality in GI contrast studies or abdominal ultrasounds. Multiple attempts to establish feeds were unsuccessful, and this included expressed breastmilk alternatives of partially hydrolyzed then extensively hydrolyzed formulas and naso-jejunal tube feeding to bypass any potential gastric or pyloric pathology, without significant improvement. Subsequently, the feeding intolerance progressed to intestinal failure with loose stool causing significant diaper dermatitis with ulceration of skin. Ultimately, his nutritional challenges were partially overcome by continuous feeds and total parental nutrition. However, he was never able to show improvement in growth parameters.

Other medical problems included urinary retention. He was unable to void spontaneously and required manual expression of the distal penis for urine to pass through the pinpoint meatus. The distal penile swelling indicated urine pooling within a urethral diverticulum. (Figure 1D). Manual urine expression was completed every 3–4 h. At 6 days of age, the patient was brought to the operative theater for an examination under anesthesia, urethral calibration, and urinary catheter insertion. The urethral diverticulum confirmed using a lacrimal probe (Figure 1C – red circle). After sequential calibration of the hypospadic pinpoint meatus, a 5 French catheter was inserted into the bladder and secured using Tagaderm. Postoperatively, he was started on trimethoprim as prophylaxis for urinary tract infection. Maintenance of the catheter was difficult due to dislodgement caused by leakage of urine around the site, which resulted in multiple reinsertions. During these multiple insertions, the tip of the catheter was left in the diverticulum, as navigation into the bladder was challenging.

Renal ultrasound ruled out hydronephrosis or congenital anomalies of the urinary system. A voiding cystourethrogram was performed with some difficulty, and the bladder was not opacified due to an inability to properly catheterize the bladder. Pooling of the contrast was seen in the urethral diverticulum (Figure 1D). The initial differential diagnosis for the genitourinary abnormality included anterior urethral valves, megalourethra, or a urethral diverticulum secondary to distal urethral atresia; all in the context of a coronal hypospadias. The working diagnosis was a coronal hypospadias with anterior urethral valves leading to a urethral diverticulum.

3 | GENETIC TESTING

3.1 | Genomic microarray analysis

Genomic microarray analysis on isolated DNA from peripheral blood cells was performed at Cytogenetics Lab of

Victoria Hospital, London Health Sciences Centre. Briefly, Infinium CytoSNP-850 K v1.2 BeadChip array (Illumina, Inc) was used for the detection of copy number variations (CNV) and loss of heterozygosity (LOH), according to manufacturer's instructions. Results were analyzed using BlueFuse Multi version 4.5(32178) (Illumina, Inc). Additional databases referenced for the analysis included DGV, gnomAD, ClinGen, ClinVar, DECIPHER, DCV, OMIM, UCSC, and CAGDB.

3.2 | Chromosomal karyotype analysis

Karyotyping was performed on trypsin-banded metaphase chromosomes with a standard protocol of 550 band resolutions from peripheral blood lymphocyte cultures. Then, ten random metaphase spreads were analyzed. Karyotypes were defined using the International System for Human Cytogenetic Nomenclature.[5]

3.3 | Fluorescence in situ hybridization (FISH)

Fluorescence in situ hybridization (FISH) assay was performed using a BAC clone RP11-454F9 probe, which maps to chromosome 17 at nucleotide position of chr17:14471748_14634927 (GRCh37/hg19 assembly). The sample subjected to standard FISH pretreatment, hybridization, and fluorescence microscopy according to the manufacturer's specifications and standard specimen-specific laboratory protocols. The results of genetic testing were described and reported in accordance with the ISCN 2016.

Furthermore, to rule out a balanced chromosomal rearrangement, involving chromosome 17, parental chromosome analysis as well as FISH analysis from the stimulated peripheral blood lymphocyte were performed.

4 | RESULTS

4.1 | Microarray result

Microarray assay exhibited a pathogenic copy number gain of approximately 21.4 Mb of chromosome region 17p13.3p11.2. between bases 8547and 21,428,752 (NCBI Build 37/hg19) (Figure 2A)., encompassing 3 established triplosensitivity regions in ClinGen Dosage Map with a score of 3 (https://dosage.clinicalgenome.org/region_search.cgi?loc=17:8547-21428752).

No other reportable copy number variants were detected, using laboratory's evaluation criteria.

4.2 | Karyotype and FISH results

4.2.1 | Proband results

Upon analysis of G-banded karyotype obtained from peripheral blood lymphocytes, a derivative chromosome 14, resulting from an unbalanced translocation between the short arm of one chromosome 14 and the short arm of one chromosome 17, was detected in all cells examined (Figure 2B). In an attempt to better characterize structural abnormalities involving chromosomes 14 and 17 observed in the G-banded karyotype analysis, additional studies were completed using FISH techniques. Following hybridization, the probe signal pattern noted was three signals (trisomic imbalance) for chromosome 17p. Two orange signals localized to anticipated chromosome 17p regions, and the third signal being localized to structurally abnormal chromosome 14, resulting in an unbalanced rearrangement (Figure 2C,D). The FISH test confirmed the characterization of the abnormalities involving chromosomes 14 and 17 that were noted in both the microarray and G-banding analyses.

4.3 | Parental results

Parental chromosome analysis and FISH showed that the mother had a normal karyotype, 46,XX, while the father carried a balanced translocation, 46,XY,t(14;17)(p11.1;p11.2).

The hybridization pattern of the FISH assay confirmed the result of chromosome analysis. (Figure 3A–C).

Incorporating the parental information and FISH results, the proband's chromosome karyotype was determined to be 46,XY,der(14)t(14;17)(p11.1;p11.2) dpat. ish der(14)(RP11-454F9+).nuc ish(RP11-454F9x3).

After genetic confirmation, and in the context of significant ongoing health co-morbidities with poor prognosis, family elected for palliative care upon several discussions with the medical care team. The proband passed on Day 70 of life at 45 weeks and 2 days postnatal age.

5 | DISCUSSION

Trisomy 17p remains a rare chromosomal disorder. It presents in a spectrum ranging from mosaics (most common), to duplications, and unbalanced translocations (as per the patient described above). Most unbalanced translocations are a result of inherited genetic material from a balanced translocation in a phenotypically normal parent,[1,3,6] and de novo occurrences have been reported[2] just the proband.

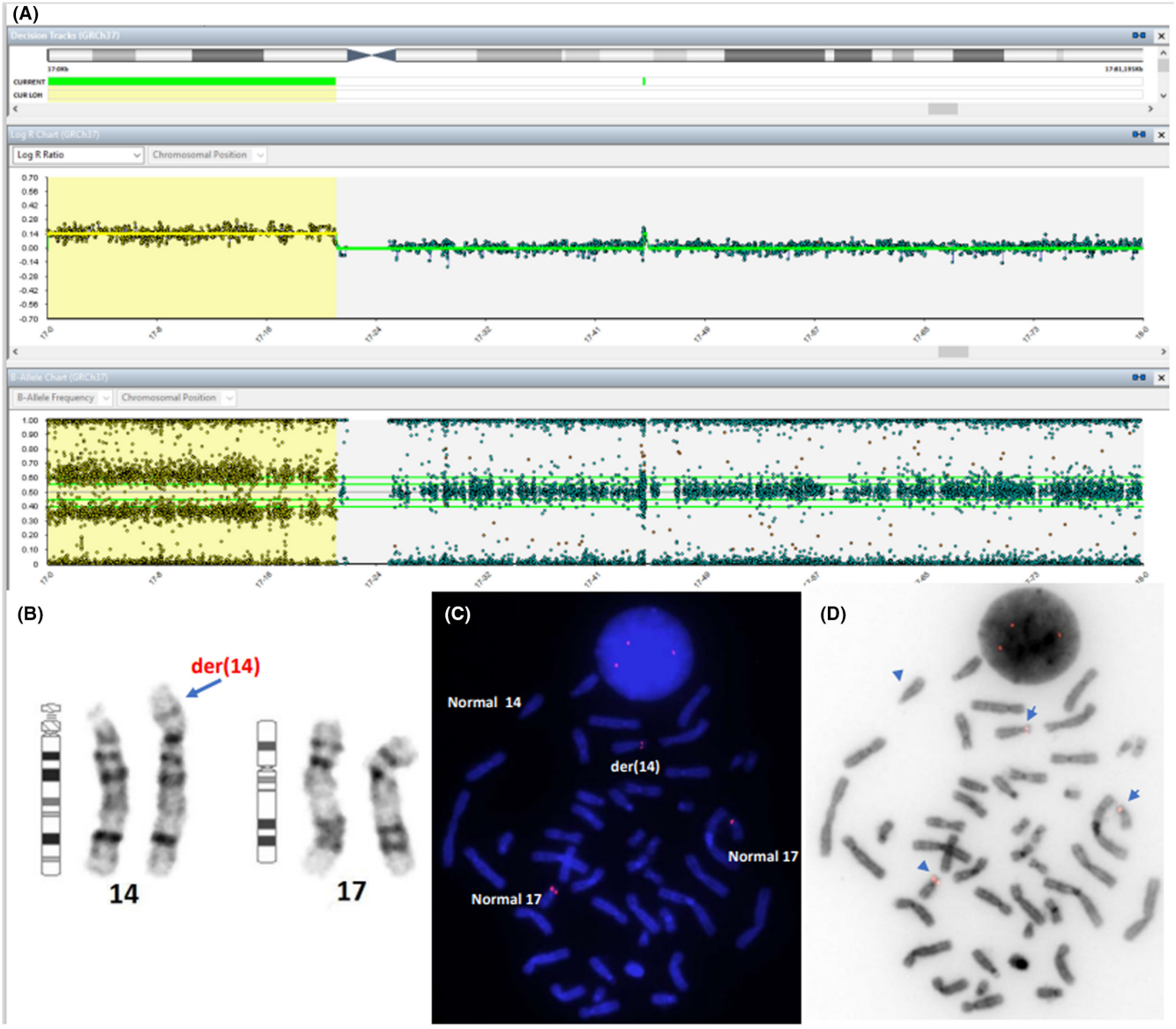

FIGURE 2 Microarray profile depicting gain (partial trisomy) of the whole short arm of chromosome 17. Representative microarray profile of the proband showing copy number state and Log2 ratio (A-top two panels) and the B-Allele Frequency (BAF) (A-lower panel) are shown for chromosome 17. (B) Peripheral blood karyotype analysis showing additional genetic material on the short arm of chromosome 14 just above the centromere (arrow). (C) Metaphase FISH analysis utilizing BAC clone RP11-454F9 probe, showing three orange signals: two on the normal 17p, and one being localized to structurally abnormal chromosome 14. The nuclei in the picture also shows three probe signals. (D) is inverted DAPI of the same metaphase

Although Trisomy 17p translocation involving chromosome 14 (as per the patient described above) has been reported earlier,[2] the proband had unbalanced translocation of paternal origin (phenotypically normal balanced translocation) and the karyotype was 46,XY,der(14)t(14;17)(p11.1;p11.2),+17. In comparison, the previously reported patient was a female and the translocation was a de novo occurrence. Table 1 illustrates the prenatal findings, karyotypes, and postnatal course of unbalanced translocation cases reported in the literature.

From genetics point of view, as it was mentioned previously, the extra genomic material from chromosome 17p in this patient contains three established triplosensitivity regions in ClinGen Dosage Map with a score of 3. The first region involves both the YWHAE (OMIM 605066) and LIS1 (OMIM 601545) genes and is associated with a variable clinical phenotype that typically includes structural brain abnormalities (involving the corpus callosum, cerebellar vermis, and cranial base), hypotonia, intellectual disability, a relatively distinct facial phenotype, and other variable findings.[7] The second region is associated with autosomal dominant Charcot–Marie-Tooth type 1A (CMT1A) (OMIM 118220). CMT1A is a neuropathy that is characterized by slowly progressive

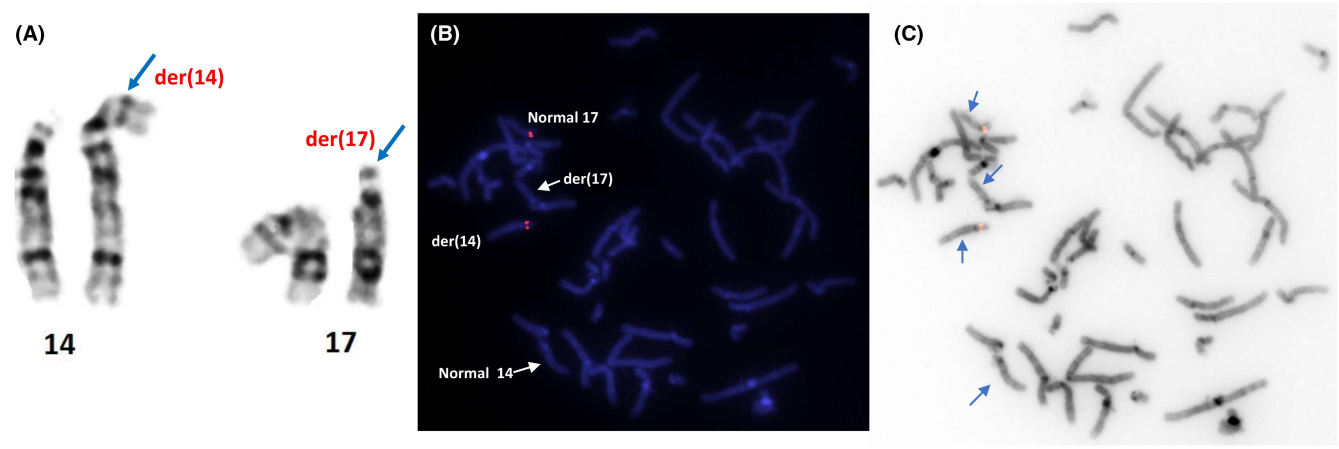

FIGURE 3 (A) Chromosome analysis from the proband's father's peripheral blood showed the balanced translocation between chromosome 14 and chromosome 17. Arrows are pointing to the translocated chromosomes. (B) Metaphase FISH analysis showing two orange signals: one on the normal 17p, and other one being localized to structurally abnormal chromosome 14. Normal chromosome 14 and derivative chromosome 17 are shown by white arrows. (C) is inverted DAPI of the same metaphase

weakness and atrophy of distal muscles (hands and the legs below the knees), hearing loss, pes cavus foot deformity, hip dysplasia, and additional clinical findings. The penetrance of this duplication is thought to be near 100%; however, age of onset and severity of the condition are variable, and some carriers are not clinically recognized. Approximately 67%–80% of recurrent 17p12 (PMP22) region duplications are inherited[8] The third region involving the RAI1 gene (OMIM 607642) is causative for Potocki-Lupski syndrome (OMIM 610883), a developmental disorder characterized by hypotonia, failure to thrive, mental retardation, pervasive developmental disorders, and congenital anomalies.[9,10]

On the contrary, prenatal diagnosis for T17 is complex as several studies have identified a phenotypically normal infant with a positive amniocentesis for T17.[11,12] Consequently, diagnostic results must be interpreted cautiously. The proband had ultrasonographic findings of IUGR prenatally and this has been reported in other case reports.[2,13] (Table 1).

Postnatally, the proband demonstrated some similarities to other cases in terms of significant dysmorphism (Figure 1A), which has been reported (Table 1), additionally he presented with an absent corpus callosum,[6] hypertonia,[1,2] feeding difficulties,[6] undescended testes,[1,3] and clinodactyly of the 5th finger.[2] In contrast, unlike other patients reported in literature, our proband did not demonstrate significant cardiac involvement.[1] However, our patient did present with unique phenotypic traits, not yet described in literature including, prolonged dependence on respiratory support and persistent feeding intolerance (without an identifiable anatomical cause for either), neurological dysfunction with increased irritability and sleep dysfunction, and genitourinary abnormality in the form of a coronal hypospadias with anterior urethral valves and associated urethral diverticulum.

This article highlights the ethical dilemma that is integral when caring for patients and families with rare genetic diagnoses. Antenatal findings were nonspecific and did not provide adequate information delaying/limiting parents' ability to assess options during pregnancy. Of the patients born alive in literature, one died at 4 months due to aspiration pneumonia after being on tube feeds,[4] there is an 8-year-old patient with no speech, hearing loss, and severe mental retardation,[2] another one is a 4-year-old patient with mild hearing loss, delayed speech, developmental delay, and feeding difficulties (requiring tube feeding).[6] In this proband, given the severity and progression of clinical disease, a shared decision-making process was implemented utilizing best available literature in conjunction with parental wishes/beliefs and a palliative approach was instituted, with withdrawal of life-sustaining therapy at DOL 68 and proband expired on DOL 70.

6 | CONCLUSION

Due to the rarity of T17p patients, clinicians face difficulty with prognostication and suggesting appropriate treatment options and goals, resulting in ethical complications. Documentation of encounters of T17p patients contributes to the database from which clinicians may draw from to care for patients and families more effectively with this genetic condition. Genetic counseling was provided to the family who were advised on the option of prenatal diagnosis in future pregnancies.

TABLE 1 Clinical characteristics of trisomy 17p unbalanced translocation cases

Author	Birth parameters karyotype	Physical, clinical, and developmental findings plus outcome
[13]	GA: 24 weeks, IUGR, right choroid plexus cyst, Amniocentesis—Trisomy 17, termination.	No autopsy
[2]	GA: 37 Weeks, IUGR BW: 1700 g 46,XX,der(14),t(14;17)(p11.2;p11.2) de novo	Dysmorphism: Triangular coarse facies, flared eyebrows, broad nasal tip with flared nares, smooth philtrum, full lips, slightly cupped low set, and posteriorly rotated ears, macroglossia, macrostomia. CNS: Reduced reflexes, hypo/hypertonia bilateral cataracts, strabismus, left eye ptosis, and hearing loss Musculoskeletal/integument: Hypertrichosis, clinodactyly of the 5th finger, scoliosis Developmental: Failure to thrive, delayed milestones (crawled at 2 years of age, walked at 5 years of age), severe mental retardation, and absence of speech.
[6]	GA: 37 Weeks BW: 1574 g (<3rd %ile) Assessed from birth to 3 years of age. 46,XX,der(22),t(17;22)(p12;p11.2) mat	Dysmorphism: Brachycephaly, broad nasal bridge, prominent eyes due to shallow orbits, down-slanting palpebral fissures, eccentric left pupil, small and angulated ears with a prominent crus on the right and rounded left ear, midline notch in upper lip, microstomia, macroglossia, flat nasal tip. CNS: Large anterior fontanelle, partial agenesis of the corpus callosum, absent rostrum and splenium, middle ear effusions (grommets inserted), and mild hearing loss. GI: Feeding difficulties (NG and G-tube feeding), high narrow palate, and anteriorly placed anus Musculoskeletal/ integument: Glabella hemangioma extending onto hooded eyelids, bilateral 5th finger brachydactyly with clinodactyly on the left and small 3rd and 4th toes and proximally inserted 5th toes. At 3 years of age increasing deformity of her right foot, walking independently using foot orthoses, weakness of shoulder girdle muscles thus weak grip. Developmental: Delayed milestones; sitting independently at 12 months, speaking 5–6 words at 18 months, crawling commando style, absent speech but able to follow simple commands using Makaton Sign Language (at 3 years of age).
[3]	GA: 38 Weeks BW: 1786 g (<10%ile) 46,XY,der(17),t(9;17)(q34.3;p13.3) mat.	Dysmorphism: Flat occiput, hypertelorism, epicanthal folds, anteverted nose, smooth philtrum, thin upper lip, low posteriorly rotated ears. CNS: Abnormal BAER, abnormal MRI (incomplete myelination), microcephaly, and decreased pigment in the ocular fundus. CVS: PPHN. GI/GU: High narrow palate, cryptorchidism, and inguinal hernias. Musculoskeletal: Bilateral clinodactyly of 5th fingers. Developmental: Developmental delay (at 4 years of age) especially in expressive language.
[1]	GA: 39 Weeks, severe oligohydramnios BW: 2570 g (<10th%ile) 46,XY,der(4)t(4;17)(q27;p11) mat	Dysmorphism: Bifid tip of the nose. CNS: Closed anterior fontanelle, narrow pupils non-responsive to light, and increased muscle tone. CVS: Complex CHD at autopsy: ASD Type II, PDA, severe preductal AS, hypoplasia of the left atrium and ventricle, dilated RA and hypertrophic RV, and absence of the sinus coronaries GU: Bilateral inguinal hernias, undescended testes (abdominal). Integument: Capillary hemangiomata over the nasal bridge and both eyelids. Outcome: Died on Day 17 from cardiorespiratory arrest.
	GA: 22 Weeks (Amniocentesis, unbalanced translocation hence termination) 46,XY,der (4)t(4;17)(p16;p11.2) pat	Musculoskeletal: Absence of 4 and 5th fingers (on Right side), deep gap between thumb and index finger, complete syndactyly between 2nd and 3rd fingers Family History: 3 Spontaneous abortions at 6–8 weeks, and one still birth at 36 weeks (male with cleft lip and palate).

(Continues)

TABLE 1 (Continued)

Author	Birth parameters karyotype	Physical, clinical, and developmental findings plus outcome
[4]	GA: 37 Weeks BW: 2050 g Assessed at birth, 2 weeks, and 3 months. 46,XX,der(8),t(8;17)(p23;p11.2)pat	Dysmorphism: Round face, hypertelorism, slanting palpebral fissures, small nose, long broad philtrum, low set ears, short, webbed neck, downward turned mouth with thin lips, and micrognathia. CNS: Dysmorphic widened ventricles. CVS: Single Umbilical Artery and PDA with cardiac murmur. GI: High arched palate Musculoskeletal/integument: Hirsutism, Short 5th fingers with clinodactyly, contractures of the elbows and fingers Developmental: SGA and Severe psychomotor retardation (At 3 months)
[14]	GA:12 Weeks Spontaneous Abortion 46,XY,-5,der(5),t(5;17)(p15.3;q23.1)mat	GI: Cleft palate Musculoskeletal: Severe bone dysplasia (Hypoplastic scapulae, short Broad clavicles, short limbs/long bones)

Abbreviations: ASD, atrioseptal defect; BAER, brainstem auditory evoked response; CHD, congenital heart disease; CNS, central nervous system; CVS, cardiovascular system; GI, gastrointestinal system; IUGR, intrauterine growth retardation; MRI, magnetic resonance imaging; PDA, patent ductus arteriosus; PPHN, persistent pulmonary hypertension of the newborn.

AUTHOR CONTRIBUTIONS

All authors were involved in the clinical management of the proband and reviewing the manuscript and approval for submission. AS, JB, and MM contributed to the initial drafting of the manuscript. MM involved in the literature search drafting of the final manuscript and submission. SB contributed to the conceptualization of the clinical case. SB, CP, PW, and AMM contributed to intellectual input in the case report.

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CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.

DATA AVAILABILITY STATEMENT

The Authors confirm that Data sharing not applicable to this article as no datasets were generated or analyzed during the current study

CONSENT

Written informed consent was obtained from the patient's parents to publish this report in accordance with the journal's patient consent policy.

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