Case Series

Understanding Patau syndrome in India: Prevalence and clinical spectrum from a tertiary care laboratory

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ABSTRACT

Trisomy 13, also known as Patau syndrome, is characterized by profound multisystem congenital anomalies and an exceptionally high rate of perinatal morbidity and mortality. A systematic case series was conducted involving four clinically suspected cases of Patau syndrome evaluated by the Department of Genetics at Nirnayan Healthcare, a tertiary care laboratory in Kolkata, West Bengal, between 2021 and 2026. After obtaining written informed consent, peripheral blood samples were collected, whole-blood cultures were established, and G-banded karyotyping was performed in strict accordance with International System for Human Cytogenomic Nomenclature 2020 guidelines to detect chromosomal abnormalities. Cytogenetic analysis confirmed trisomy 13 in all four cases, presenting in different cytogenetic forms: Pure trisomy with karyotypes 47,XY,+13 and 47,XX,+13; mosaic trisomy represented as 47,XY,+13[20]/46,XY[30]; and a Robertsonian translocation involving two chromosomes 13, designated as 46,XX,+13,der(13;13)(q10;q10). This case series demonstrates the cytogenetic diversity of Patau syndrome in Eastern India, including full trisomy, mosaicism, and Robertsonian translocation, underscoring the importance of accurate cytogenetic diagnosis for effective clinical management and genetic counseling. It also highlights significant gaps in access to genetic services, particularly in rural areas, leading to delayed diagnosis and increased familial burden. Expanding prenatal screening programs and integrating genetic services into public health systems are essential for early detection, informed reproductive decision-making, and improved clinical outcomes.

Key words: Genetic anomalies, Mosaic Patau syndrome, Robertsonian translocation, Trisomy 13

Patau syndrome (trisomy 13), caused by the presence of an additional copy of chromosome 13, was first described by Klaus Patau in 1960 [1]. It is characterized by multiple severe congenital anomalies, including congenital heart defects, postaxial polydactyly, ocular abnormalities, hypotonia, cleft lip and palate, and renal malformations, with considerable variability in clinical severity among affected individuals [2]. Although rare, occurring in approximately 1 in 12,000–20,000 live births worldwide, trisomy 13 is associated with an extremely poor prognosis, with most affected pregnancies resulting in spontaneous abortion or death within the 1st month of life [3,4]. Cytogenetic analysis remains the gold standard for definitive diagnosis.

Despite its significant clinical burden, population-based data on the prevalence, clinical spectrum, and cytogenetic mechanisms of Patau syndrome remain scarce in India. In settings where access to prenatal screening and specialized genetic services is restricted, particularly in rural and resource-constrained regions, many cases are diagnosed only after birth. This delayed recognition often deprives families of timely counseling and informed reproductive decision-making, underscoring the importance of cytogenetic evaluation in tertiary care centers. Patau syndrome arises primarily from meiotic non-disjunction, resulting in an extra chromosome 13, and occurs in three principal cytogenetic forms: Trisomy 13, mosaicism, and translocation trisomy 13 [5]. The expression and severity of congenital anomalies are influenced by multiple factors, including maternal age, consanguinity, and the specific chromosomal mechanism involved [6]. In India, where consanguineous marriages are common in certain populations and access to genetic screening is uneven, understanding these cytogenetic patterns is of particular clinical and public health relevance.

In this case series, we systematically analyzed four cases of Patau syndrome diagnosed at a tertiary care

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genetics laboratory in Eastern India. By correlating detailed clinical features with cytogenetic findings, we aimed to characterize the spectrum of phenotypic manifestations and underlying chromosomal mechanisms observed in this setting. This case series seeks to contribute region-specific data on trisomy 13, highlight diagnostic challenges, and emphasize the critical role of early cytogenetic testing and genetic counseling for affected families.

CASE SERIES

A systematic case study was undertaken involving four clinically suspected cases of Patau syndrome evaluated by the Department of Genetics at Nirnayan Healthcare, a tertiary care diagnostic laboratory located in Kolkata, West Bengal, over a 5-year period from 2021 to 2026. Nirnayan Healthcare is duly registered under the Department of Health and Family Welfare, Government of West Bengal, and licensed in accordance with the Clinical Establishments Act, 2017 (License No. 33735154). Ethical approval for the study was obtained before commencement. Detailed clinical histories were obtained for each patient, and comprehensive phenotypic assessments were performed based on information extracted from physicians' prescriptions, referral notes, and hospital discharge summaries. These records were carefully reviewed to document key clinical features, congenital anomalies, and developmental abnormalities suggestive of trisomy 13.

Following the acquisition of written informed consent from the patients' guardians in accordance with ethical standards, peripheral venous blood samples were collected under aseptic conditions. Whole-blood cultures were established using standard lymphocyte culture techniques, with appropriate mitogenic stimulation to promote cell division. After incubation, cells were harvested, treated with a hypotonic solution, and fixed to obtain well-spread metaphase chromosomes. Giemsa banding (G-banding) was subsequently performed to enable high-resolution visualization of chromosomal banding patterns.

Chromosomal analysis was carried out following the guidelines set forth by the International System for Human Cytogenomic Nomenclature 2020. A minimum of 20 metaphase spreads per case were examined to ensure diagnostic accuracy, with additional cells analyzed in cases where mosaicism was suspected. This approach facilitated the detection and characterization of numerical and structural chromosomal abnormalities, including full trisomy 13, mosaic variants, or translocation-associated forms of Patau syndrome. The cytogenetic findings were then correlated with the observed clinical phenotypes to strengthen genotype–phenotype associations and to support definitive diagnosis.

Four clinically suspected cases of Patau syndrome demonstrated considerable phenotypic variability, reflecting different cytogenetic mechanisms, including full trisomy 13, mosaic trisomy 13, and translocation-associated trisomy 13. The salient clinical features and cytogenetic findings of each case are described below.

Case 1

A 2-day-old female neonate was referred for cytogenetic evaluation due to multiple congenital anomalies. She was born to non-consanguineous parents with a history of advanced maternal age. The birth weight was 1.8 kg. The antenatal history was unremarkable, with no reported maternal illness or exposure to teratogenic agents during pregnancy. Parental karyotypes were normal (46,XX in the mother and 46,XY in the father).

Clinical examination revealed craniofacial and central nervous system anomalies, including microcephaly and holoprosencephaly. Limb anomalies included postaxial polydactyly. Additional findings were cleft palate, congenital heart disease, generalized hypotonia, and renal abnormalities detected on imaging. However, certain features commonly associated with Patau syndrome, such as microphthalmia, cutis aplasia, and low-set ears, were absent. The involvement of multiple organ systems raised a strong clinical suspicion of trisomy 13. Chromosomal analysis demonstrated a karyotype of 47,XX,+13, confirming the diagnosis of trisomy 13 (Patau syndrome). The cytogenetic findings were consistent with the observed clinical phenotype. Genetic counseling was provided to the family regarding prognosis and recurrence risk in future pregnancies.

Case 2

A 1-week-old male neonate was referred for evaluation of multiple congenital anomalies. He was born to consanguineous parents, with no history of advanced maternal age, and had a birth weight of 1.6 kg. The mother had a history of one prior pregnancy loss. Cytogenetic analysis of the parents showed a normal karyotype in the father (46,XY), while the mother carried a common heterochromatic variant, 46,XX,9qh+, which is considered a benign polymorphism with no known clinical significance.

Clinical examination revealed prominent craniofacial and central nervous system abnormalities, including microcephaly, holoprosencephaly, microphthalmia, cutis aplasia, and low-set ears. A cleft palate was present. Cardiovascular evaluation identified congenital heart disease, and the neonate exhibited generalized hypotonia. Polydactyly was absent, and no renal abnormalities were detected on clinical or radiological assessment. The constellation of severe craniofacial anomalies and central nervous system malformations strongly supported a diagnosis of trisomy 13. Cytogenetic analysis of peripheral blood revealed a karyotype of 47,XY,+13, confirming trisomy 13. The extensive phenotypic abnormalities showed a strong correlation with the cytogenetic findings. Post-diagnostic genetic counseling addressed the genetic

etiology, expected clinical outcome, and recurrence risk, particularly in the context of parental consanguinity.

Case 3

A 5-month-old male infant, weighing 4.5 kg, was referred for genetic evaluation due to developmental delay and multiple congenital anomalies. He was born to non-consanguineous parents with a documented history of advanced maternal age. No clear antenatal history of maternal illness or teratogenic exposure was available. Parental karyotypes were normal (46,XX and 46,XY).

Clinical examination showed comparatively milder features, including postaxial polydactyly, generalized hypotonia, and microphthalmia. Renal abnormalities were detected on radiological assessment. In contrast, several major features commonly associated with Patau syndrome – such as cleft palate, congenital heart disease, microcephaly, cutis aplasia, holoprosencephaly, and low-set ears – were absent. The variable phenotype and survival beyond the neonatal period suggested the possibility of chromosomal mosaicism. Cytogenetic analysis revealed a mosaic karyotype of 47,XY,+13[20]/46,XY[30], indicating the presence of two cell lines: One with trisomy 13 and one with a normal male chromosomal complement. This confirmed the diagnosis of mosaic trisomy 13. The mosaic pattern correlated with the less severe clinical presentation and prolonged survival. Genetic counseling emphasized the typically sporadic nature of mosaic trisomy 13 and discussed options for prenatal diagnosis in future pregnancies.

Case 4

A 5-day-old female neonate was referred following the identification of multiple congenital anomalies. She was born with a birth weight of 1.9 kg to consanguineous parents, with no history of advanced maternal age. The mother had a history of three spontaneous abortions (P0+3).

Clinical evaluation revealed craniofacial and central nervous system anomalies, including microphthalmia, cutis aplasia, holoprosencephaly, and low-set ears. A cleft palate was present. Cardiovascular assessment identified congenital heart disease, and renal abnormalities were detected on imaging. The neonate also exhibited generalized hypotonia. Microcephaly and polydactyly were absent. The combination of characteristic facial anomalies and multisystem involvement strongly suggested Patau syndrome. Cytogenetic analysis showed a karyotype of 46,XX,+13,der(13;13)(q10;q10), consistent with translocation-associated trisomy 13 due to an unbalanced Robertsonian translocation between two chromosome 13 homologs. Further parental karyotyping revealed paternal inheritance of the balanced Robertsonian translocation, with the father having a karyotype of 45,XY,der(13;13)(q10;q10), while the mother had a normal karyotype (46,XX). These findings confirmed the diagnosis of Patau syndrome resulting from a structural chromosomal rearrangement

Table 1 summarizes the key clinical characteristics of the four cases, including age, sex, body weight, consanguinity, and documented congenital anomalies at presentation, and Figure 1 shows the karyotypic analysis

Table 1: Comparative summary of clinical features, cytogenetic findings, and parental background in four cases of Patau syndrome

Parameter	Case 1	Case 2	Case 3	Case 4
Sex/age at presentation	Female/2 days	Male/1 week	Male/5 months	Female/5 days
Body weight (kg)	1.8	1.6	4.5	1.9
Parental consanguinity	No	Yes	No	Yes
Advanced maternal age	Yes	No	Yes	No
Obstetric history	Normal	One prior pregnancy loss	Normal	Three spontaneous abortions (P0+3)
Antenatal history	Unremarkable	Unremarkable	Not clearly documented	Unremarkable
Craniofacial/CNS anomalies	Microcephaly, holoprosencephaly	Microcephaly, holoprosencephaly	Absent	Holoprosencephaly
Microphthalmia	Absent	Present	Present	Present
Cutis aplasia	Absent	Present	Absent	Present
Cleft palate	Present	Present	Absent	Present
Low-set ears	Absent	Present	Absent	Present
Polydactyly	Present (postaxial)	Absent	Present (postaxial)	Absent
Congenital heart disease	Present	Present	Absent	Present
Generalized hypotonia	Present	Present	Present	Present
Renal anomalies	Present	Absent	Present	Present
Clinical severity	Severe	Severe	Mild to moderate	Severe
Cytogenetic finding	47,XX,+13	47,XY,+13	47,XY,+13[20]/46,XY[30]	46,XX,+13,der(13;13)(q10;q10)
Parental karyotype	Normal	Mother: 46,XX,9qh+	Normal	Father: 45,XY,der(13;13)(q10;q10)

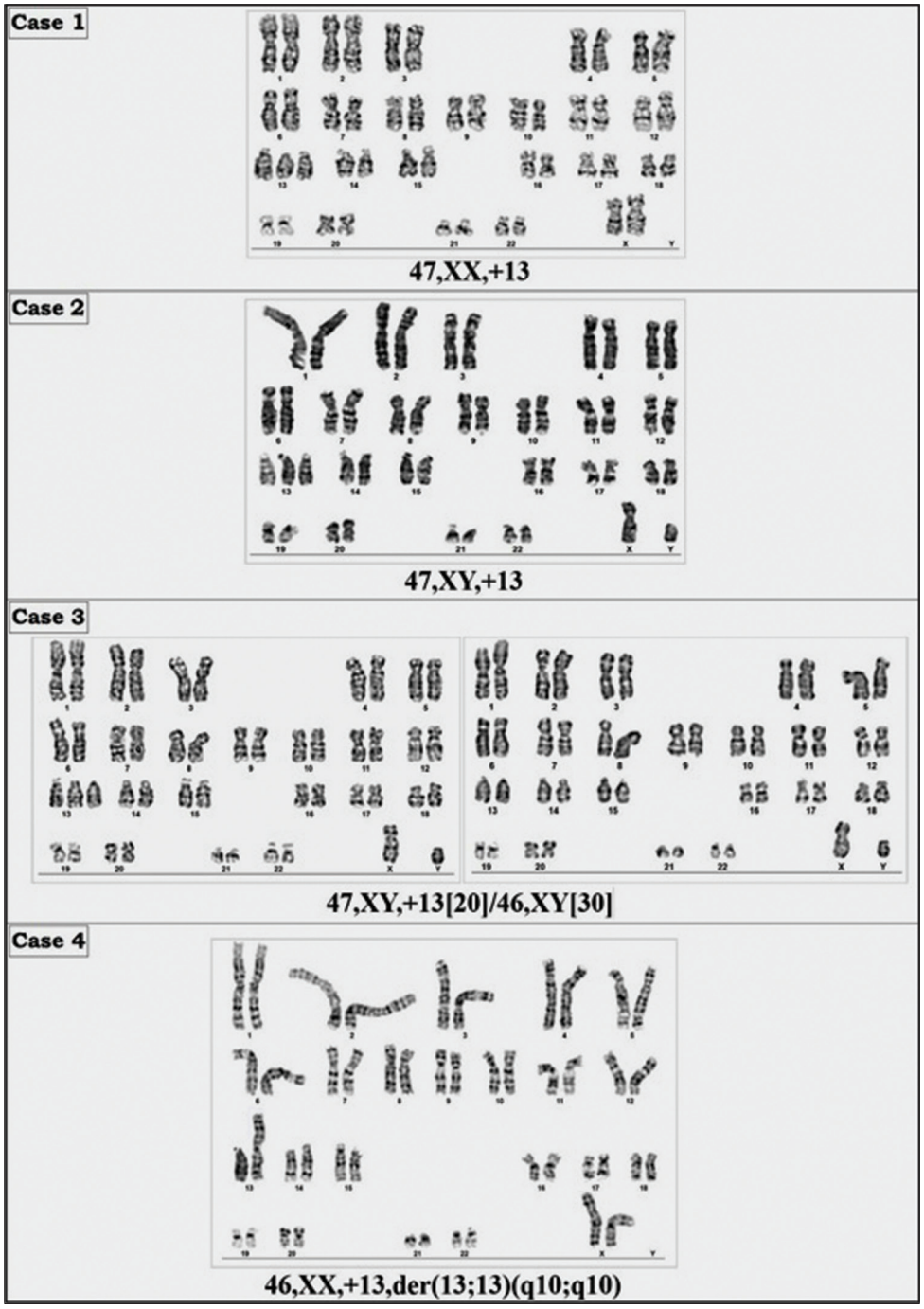

Figure 1: The observed karyotyped chromosomal analysis with trisomy 13 in all four cases, with distinct cytogenetic variants, including complete trisomy in case 1(47,XY,+13) and case 2 (47,XX,+13), mosaic trisomy in case 3 (47,XY,+13[20]/46,XY[30]), and a Robertsonian translocation between two chromosomes 13 in case 4 designated as 46,XX,+13,der(13;13)(q10;q10)

demonstrating trisomy 13 in all four cases, with distinct cytogenetic variants.

DISCUSSION

Patau syndrome (trisomy 13) is a rare and severe chromosomal aneuploidy characterized by multisystem congenital anomalies and high neonatal mortality [3-6]. The present case series highlights the wide phenotypic spectrum and distinct cytogenetic mechanisms underlying trisomy 13, including full trisomy, mosaicism, and translocation-associated forms [5,7]. Although all four patients were clinically suspected of Patau syndrome, their presentations varied considerably.

Trisomy 13 has an estimated incidence of approximately 1 in 12,000–20,000 live births worldwide and is associated with a high rate of antenatal loss and early postnatal death [3-6]. A majority of cases are caused by complete (homogeneous) trisomy due to meiotic non-disjunction, while Robertsonian translocations and mosaicism account for smaller proportions of cases [5]. International literature documents several atypical and informative presentations of Patau syndrome. Rare reports of long-term survival in individuals with mosaic trisomy due to mechanisms such as incomplete trisomic rescue illustrate the complex genetic dynamics that modulate phenotype and survival [7-10]. A patient with mosaic proximal trisomy 13q and a minor free trisomy 13 cell line exhibiting relatively extended survival with

complex phenotypic features has been described [10]. Another report detailed a child with mosaic trisomy 13, highlighting the potential for prolonged life span and variable clinical features in mosaic cases [8]. These cases underscore the need for nuanced clinical evaluation and the recognition that mosaicism and structural variants may contribute to atypical survival trajectories.

In India, trisomy 13 has been reported mainly through isolated case reports and small series, reflecting both its rarity and the limited availability of large-scale cytogenetic and epidemiological data [11-17]. Several Indian case reports describe clinical diagnoses confirmed by karyotyping, with phenotypes ranging from classical presentations to milder and atypical forms [11-17]. For example, an antenatal diagnosis based on ultrasonography and subsequent karyotyping in Andhra Pradesh highlighted typical anomalies, including polydactyly and midline defects [13]. Another Indian report described a milder and later presentation of trisomy 13 with extended survival beyond infancy, which is uncommon in full trisomy [14]. Case reports from regions such as Jammu and Kashmir and Karnataka further support the phenotypic and cytogenetic variability seen in Indian neonates [15,16]. Despite these reports, comprehensive, large-scale epidemiological data on trisomy 13 in India are limited, emphasizing the need for broader cytogenetic surveillance and reporting efforts.

Two of the cases in this series (Cases 1 and 2) demonstrated full trisomy 13 (47,XX,+13 and 47,XY,+13), the most common form of the disorder [5]. These patients exhibited multiple congenital anomalies involving craniofacial structures, cardiac defects, and central nervous system malformations, consistent with classical descriptions in international cohorts [2,3,6]. Case 2, in particular, presented with extensive holoprosencephaly, cutis aplasia, and microphthalmia, illustrating the phenotypic severity often seen in complete trisomy 13. Conversely, Case 1 lacked several classical features such as microcephaly and microphthalmia, demonstrating that even within full trisomy, expressivity can be variable. Such phenotypic heterogeneity has been documented in retrospective analyses reporting variable clinical manifestations among cytogenetically confirmed trisomy 13 patients [6].

Advanced maternal age is a recognized risk factor for meiotic non-disjunction leading to autosomal trisomies, an association observed in Cases 1 and 3, although Case 2 demonstrates that trisomy 13 can also arise sporadically [11]. Case 3 demonstrated mosaic trisomy 13 (47,XY,+13[20]/46,XY[30]), a less common form resulting from postzygotic mitotic errors [8,9]. Mosaicism can lead to a mixture of normal and trisomic cell lines, often resulting in milder phenotypes and extended survival compared to full trisomy [8-19]. The considerably variable clinical outcomes in mosaic cases have been reflected in reports of long-term survivors who exhibited relatively milder anomalies but persisted with developmental challenges [8,17]. Case 4 represented translocation-associated trisomy 13, caused by an unbalanced Robertsonian translocation between two chromosome 13 [5,18]. Robertsonian translocations represent a structural chromosomal mechanism in which the long arms of two acrocentric chromosomes fuse, potentially leading to partial or complete trisomy of the involved chromosome [5]. The identification of an unbalanced Robertsonian translocation has significant implications for genetic counseling, as one parent may be a phenotypically normal balanced carrier, substantially increasing recurrence risk; therefore, parental karyotyping is essential to guide reproductive planning [18,19]. In this case, the der (13;13) chromosome was found to be paternally inherited. Robertsonian translocation between two homologous chromosomes is unique in that carrier parent of these rearrangements will by necessity pass the abnormal chromosome to all offsprings making the recurrence risk of translocation Patau syndrome 100% [18,19]. Accordingly, parental karyotyping was recommended to determine carrier status, and the family was counseled regarding recurrence risk, options for prenatal diagnosis, and the availability of preimplantation genetic testing in subsequent pregnancies.

Collectively, this case series demonstrates a clear genotype–phenotype correlation in Patau syndrome: Full trisomy often leads to severe multisystem anomalies and early mortality, mosaicism is associated with variable expressivity and improved survival prospects, and translocation-associated trisomy has significant hereditary implications requiring specialized counseling [5,6,8-10,17-19]. The variability observed across cases reinforces that reliance on clinical features alone is insufficient for definitive diagnosis, emphasizing the essential role of cytogenetic analysis in accurate diagnosis, prognosis, and family counseling.

CONCLUSION

Comprehensive cytogenetic evaluation using G-banded karyotyping remains indispensable in suspected cases of Patau syndrome, particularly in the Indian healthcare context, where genetic services are unevenly distributed. Accurate identification of the underlying chromosomal mechanism is crucial for prognosis, management, and informed genetic counseling. Early diagnosis enables targeted reproductive planning through prenatal testing and, when appropriate, preimplantation genetic testing, thereby reducing the risk of recurrence in affected families.

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