International Journal of Medical Research Professionals
P-ISSN: 2454-6356; E-ISSN: 2454-6364
DOI: 10.21276/ijmrp

IJMRP

Case Report .

Patau Syndrome Survival 6 Years & It's Associated Ocular Findings: Case Report

Aisha Mohammed Alemam[1*], Abdulrahman Alturki[1], Abdulqader AlHebshi[2], Sami Alloqmani[3]

[1]Department of Medicine, Taibah University Faculty of Medicine, Al Madinah Al Munawwarah, KSA.
[2]Department of Pediatrics, Prince Mohammad bin Abdulaziz Hospital, Almadinah, KSA.
[3]Pediatric Resident, Maternal Child Hospital, Madinah, KSA.

ABSTRACT

Trisomy 13 is a rare syndrome with a prevalence of 1:12000-1:29000 in newborns. Eighty-two percent die within one month. The majority of patients with patau syndrome have several ophthalmic abnormalities including microphthalmos, cyclopean synophthalmus, inferonasal iris clobomas of the uveal tract, cataracts and persistent hyperplasia of primary vitreous body and those finding represent 80% of cases. A 6-year-old and child was born at full term with microcephaly and CNS abnormalities. Late onset of seizures started when she was 2 years old, ASD, PDA, cleft lip and palate, microphthalmia, inferonasal iris cloboma and retinal detachment. Comparing it with 13 other cases of long survival patau syndrome to investigate the causes of longevity and to study it's associated ocular anomalies. factors associated with prolonged survival rate are not fully understood, but many studies have mentioned that certain factors including genetic types translocations or mosaicism, race, female gender increase the survival rate. Most ophthalmic manifestations associate with patau syndrome are microphthamia, cloboma and cataract. We are reporting a 6 years old girl with patau syndrome studying it's ocular findings and we have done a literature review to investigate the causes associated with long term survival since patau syndrome has poor vital prognosis.

Keywords: Genetic; Patau; Trisomy 13; Ocular.

***Correspondence to:**

Aisha Mohammed Alemam,
Department of Medicine,
Taibah University Faculty of Medicine,
Al Madinah Al Munawwarah, KSA.

Article History:

Received: 23-09-2017, **Revised:** 18-10-2017, **Accepted:** 24-12-2017

Access this article online	
Website: www.ijmrp.com	Quick Response code
DOI: 10.21276/ijmrp.2018.4.1.063	

INTRODUCTION

Patau Syndrome (Trisomy 13) was first identified as a genetic disorder in 1960 thats caused by an extra copy of chromosome number 13.[1] Most cases die before the age of 6 months from severe abnormalities in the heart, kidney and brain failure which is associated with developmental delay. Eighty-two percent die within one month[2] and eighty-five percent do not live beyond one year.[3] Survival beyond one year of age was found to be associated with mosaicism.[4] The syndrome is accompanied by multiple malformations including facial malformations as cleft palate and lip, skeletal malformation as polydactyly and clubfoot, CNS malformation, heart defects and abnormalities of the genital tract.[5] Facial clefting can be associated with other midline defects, which may adversely affect the survival rate of an individual.[6] The majority of patients with patau syndrome have severe ophthalmic abnormalities, the most frequent and typical malformations are microphthalmos, cyclopean synophthalmus, inferonasal iris clobomas of the uveal tract, cataracts and persistent hyperplasia of primary vitreous body and those findings represent 80% of cases.[7,8] 75% have dysplasia of the retina, 60% have anterior chamber dysgenesis associated with glaucoma and 65% have cartilage within coloboma.[8]

In this study we report a case of a female patient who's 6 years old in age diagnosed with full trisomy 13 (47, XX + 13) who has survived longer than usual and studying it's associated ophthalmic manifestations and other anomalies.

CASE REPORT

A 6-year-old Saudi girl, a fifth girl of a non-consanguineous couple. Mother and father both aged 40, previously had 4 healthy children, G5 P4+0, and had a past history of myomectomy. All previous children were healthy and delivered by spontaneous vaginal delivery except the second baby, had intra uterine growth retardation. The pregnancy was monitored but not regularly and was uneventful. At the first month of pregnancy, sonography was done Figure (1).

The fetal pole was seen with positive cardiac pulsation without internal os dilated. No scanning for fetal anomalies or alpha fetoprotein level were done during pregnancy.

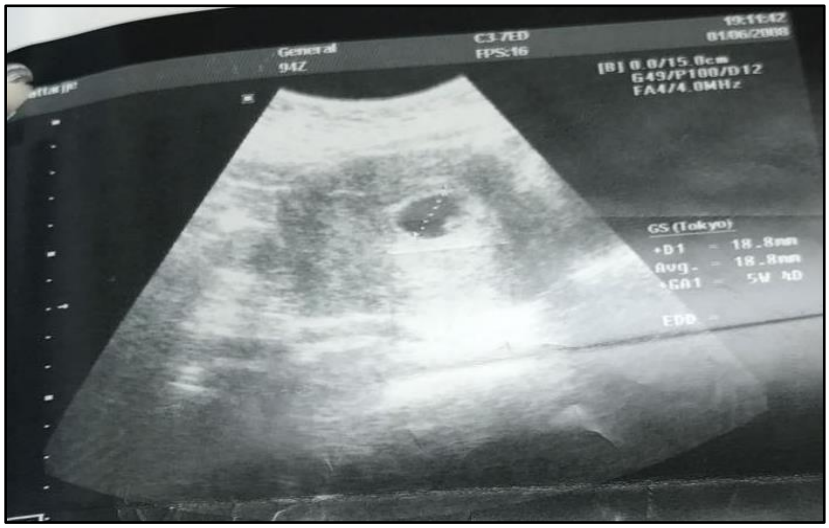

Fig 1: Sonography showed Intrauterine gestational salk measuring 19 mm that's equal 5 weeks and 5 days

Table 1: APGAR Score

System	0	1	2	1 min	2 min	3 min
Respiratory effort	**Absent**	**Slow irregular**	**Good crying**	0	1	1
Heart rate	**Absent**	**Below 100**	**Over 100**	1	2	2
Color	**Blue-pale**	**Body pink blue extremities**	**Completely pink**	0	2	2
Muscle tone	**Limp**	**Somellexion of extremities**	**Active motion**	1	1	2
Reflex irritability	**No. response**	**Grimace**	**Cry**	1	1	1
Total				3	7	8

In the third trimester, the mother had pregnancy induced hypertension, which planed her for a caesarian section. No drug history even supplementation of iron and folic acid and she was not exposed to radiation. The child was born at full term 37 weeks of pregnancy delivered by caesarean section. Apgar score was 3 in 1 minute, 7 in 5 minutes and 8 in 10 minutes (Table 1).

No meconium aspiration. Birth weight was 2.5Kg, length was 49cm and head circumference was 33cm. The initial examination at birth showed dysmorphic features consistent with patau syndrome, unilateral cleft lip and bilateral cleft palate, low set ears, hypertelorism, midfacial hypoplasia, microcephaly, absent teeth, ASD secondum lift to right and PDA, bilateral microphthalmia, inferonasal iris clobomata. The child was hypotonic and jaundiced and was admitted to the NICU at the day of delivery and stayed for 1 month. Jaundice was managed by phototherapy. The child was kept on ventilation, blood culture was done and showed gram positive cocci, and she received a full course of antibiotics, blood and fresh frozen plasma transfusion.

At the age of 1 month, the child was referred to genetic counseling. Cytogenetic preliminary study was done and the diagnosis of Patau syndrome (trisomy 13) was proved by chromosomal study with a karyotype 47, XX + 13. The family was informed about the disease. Lip repair and vomer flap was done when the child was 4 months and 20 days old. The cardiac pathology was followed by a cardiologist till large ASD secondum left to right shunt, then became small and PDA confirmed by echocardiogram and fortunately closed at 7 months in 14/7/2009.

In ophthalmology review she was found to have an inferonasal iris clobomata. A-scan examination of the right and left eye was done showing an anteroposterior axis of 9.6, 9.9 mm respectively confirming the diagnosis of bilateral microphthalmia, she was examined under GA when she was 3 months old, found to have telangectic vessles inferonasally with serous RD OS, serial cryotherapy sessions were done till examination under GA showed no active telangiectasia and resolved subretinal fluid.

When she was 2 years old, she was admitted in 2/2/2010 complaining of cough, diagnosed as bronchial pneumonia and managed by vancomyocin with imipenem. During this admission, she was diagnosed with epilepsy according to the EEG findings, which showed: abnormal EEG stand revealing generalized epileptic discharge, and treatment with Phenobarbital was started.

At the age of 2 years, she started to attend physiotherapy sessions. Motor and intellectual development were delayed for her age. Hearing Examination showed decrease in hearing bilaterally.

Child was followed up in ophthalmology, cardiac, psychomotor developmental and physiotherapy clinics with no history of admission until 5 years. At the age of 5 years, in 16/06/2014 the patient was admitted to PMBAH to do a brain CT (Fig 2) under Chloral Hydrate, because she had developmental delay.

She had recurrent admissions due to respiratory infection. In 19/06/2015 the patient was admitted for cough and was diagnosed with gastro-esophageal reflux disease and associated aspiration pneumonia. Chest X-ray was done (Fig 3) .She was on oxygen by nasal cannula, lorazepam, sodium valproic acid and augmentin then she was shifted to PICU and was on phenobarbital. She was discharged in 1/07/2015. In 25/07/2015 the patient was admitted for cough and fever for 1 day and she was diagnosed with an URTI. An X-ray was done showing (fig 4). She was planned to be on ceftroxime and gentamycin and was discharged on 22/07/2015.

In 4/10/2015, she developed apnea while she was sleeping and died at age of 6 years and 6 months.

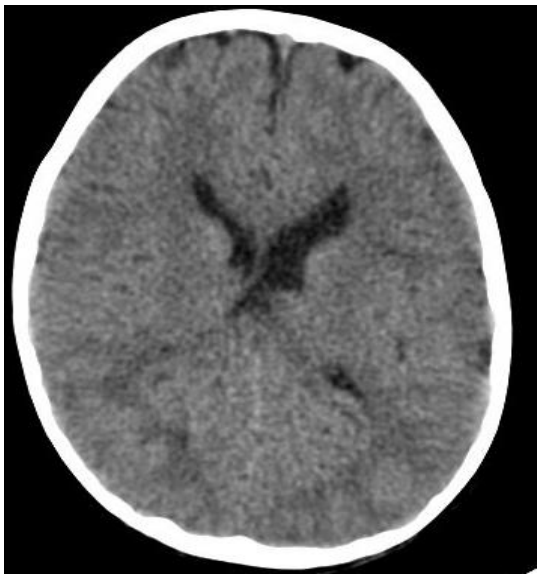

Fig 2: CT Scan showed: evidence of asymmetry of the cerebral hemispheres. The left one was slightly smaller. There was also asymmetry of the ventricular system. The left lateral ventricle was larger, most likely due to some atrophy of the left cerebral hemisphere. Bilateral symmetrical globus palidum calcifications are noted. enlarged optochiasmatic cistern and peri pontine cistern with some atrophy of optic chiasma are noted. There was also hypoplasia of the splenium and posterior part of corpus callosum. Eyes globes were unremarkable. There was no evidence of cranio stenosis.

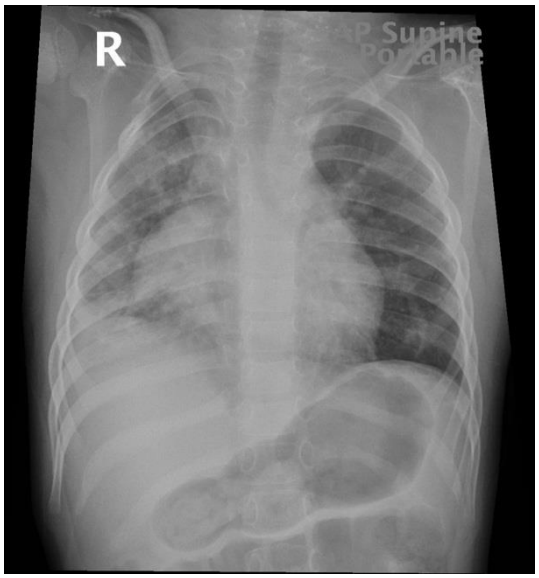

Fig 3: Chest X-ray showing: evidence of bilateral parahilar congestion, obliteration of the right CP angle, left CP angle was patent and the bony cage is unremarkable with mild pleural effusion on the right side. Cardiac size was not well assessed, but there may be mild cardiomegaly.

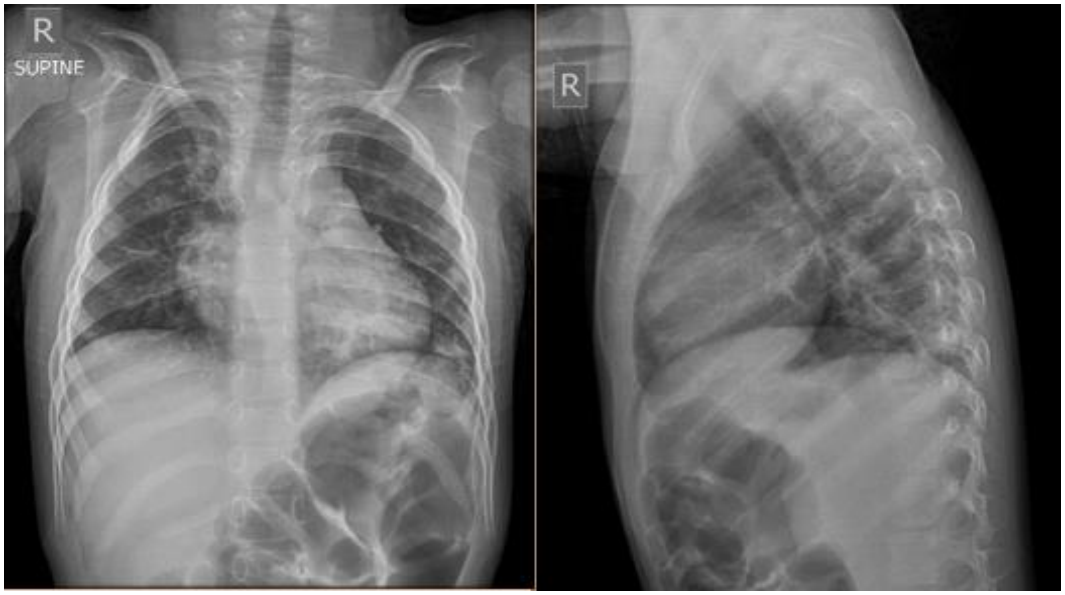

Fig 4: Chest X-ray showing: The heart was enlarged and this might be due to supine and AP view. Consolidation was seen at the left lower lobe in the perihilar area suggestive of infection, for clinical correlation and follow up after antibiotic therapy with no signs of pleural effusion or pneumothorax

DISCUSSION

Trisomy 13, is a genetic disorder associated with low survival rate among infant and high rates of spontaneous abortion. 85-90% of cases died during the first year of life; only 5% to 10% of patients live beyond the age of one year, and patients who survive beyond one year have a sever developmental handicap.[9] Factors associated with prolonged survival rate are not fully understood, but many studies have mentioned that certain factors including genetic types translocations or mosaicism, race, female gender increase the survival rate.[10] In this study we are reporting a case that lived for 6 years and 6 months who had microcephaly and CNS abnormalities, late onset of seizure started when she was 2 years old, ASD, PDA, cleft lip and palate, microphthalmia, ifneronasal iris cloboma and retinal detachment. Comparing it with 13 other cases of long survival patau syndrome to investigate the causes of longevity and to study it's associated ocular anomalies. From the literature we found that holoprosencephaly, early onset of seizure before age of 1year, scalp defect, VSD, hydronephrosis and kidney agenesis were not or were rarely seen in long survival cases. The number and severity of anomalies often directly correlates with survival.[11] Therefore, the clinical findings of trisomy 13 are the most important factors in prolonged survival rate. We

believe that non-lethal and comprehensive medical care play a role in long survival rate among patients with trisomy 13.

Since patau syndrome is often a lethal condition in infancy and early childhood almost all reported ocular abnormalities based on histopathological studies on autopsy specimens, only a few reports describes the clinical ocular abnormalities in live patients.[12,13] Studies showed 88% of cases with trisomy 13 have ocular abnormalities.[14] The definitive cause of ocular abnormalities in patau syndrome is unknown, but it's related to chromosomal aberration. This case has bilateral inferonasal iris clobomatas, microphthalmia and inferonasal retinal telangiectasia with retinal detachment in the left eye.

According to our literature review the most common ocular abnormalities were microphthamia, cloboma and cataract which mimic the result of yanoff, fayer and scheie' study.[15] Other ocular abnormalities were reported in several studies, corneal opacification was reported by smith et al in 1963[16], and presence of intraocual cartilage was noticed by Cogan and Kuwabara in 1964.[17] Cloudy cornea and buphthalmos was reported by keith.[18] Magni reported a case with iris and retinal clobomas, ectopic lentis and clobomatous orbital cyst.[19]

According to our literature review corneal, conjunctival and extra ocular muscles anomalies were not frequent in trisomy 13 while anterior uveal tract, lens optic nerve hypoplasia, angle structure anomalies and retinal anomalies were frequently seen. Although ophthalmic manifestations alone are not sufficient to reach a definitive diagnosis of patau syndrome, any child presented with cataract and inferonasal iris clobomas with dysmorphic features should have karyotype testing to rule out trisomy 13.[20]

One third of patau sundrome patients were born prematurely, so it's important to differentiate the pathological ocular finding of patau syndrome from those related to prematurity. For example, corneal stromal hypercellularity and incomplete angle cleavage might be caused by prematurity, however retinal dysplagia and inferonasal iris clobomatas are pathological.

CONCLUSIONS

We are reporting a 6 years old girl with patau syndrome studying it's ocular finding (bilateral microphthalmia, inferonasal iris clobomata) and we have done a literature review to investigate the causes associated with long term survival since patau syndrome have poor vital prognosis, 85% of patients die within 1 year of age. There has been a number of reports on patau syndrome patients showing mosaic genotyping and the number and severity of malformation have significant contribution on long term survival.

We hope this report contributes to further understanding of the factors associated with longevity in survival to improve the medical care services provided to this group and to improve the vital prognosis.

ACKNOWLEDGMENT

Authors gratefully acknowledge Hissah Abdullah Alturki, Abeer Abdulaziz Alradadi and Malik Jelaidan for support and help us.

REFERENCES

1. Patau K, Smith DW, Therman E, et al. Multiple congenital anomaly caused by an extra chromosome. Lancet 1960;1:790.

2. Best, R.G. (2002). Patau syndrome. On-line article: www.emedicine.com/PED/topic1745.htm. Accessed Jan 14, 2003

3. Widerемann, H.R., Grosse, F.R. and Dibben, H. (1980). Caracteri□stica das Si□ndromes em Pediatria. Editora Manole Ltda., Sa□o Paulo, SP, Brazil.

4. Hsu HF, Hou JW. Variable expressivity in Patau syndrome is not all related to trisomy 13 mosaicism. Am J Med Genet A 2007;143A:1739–48.

5. Taylor AI. Autosomal trisomy cases: a detailed study of 27 cases of Edward's syndrome and 27 cases of Patau's syndrome. J Med Genet 1968;5:227.

6. Koury MJ, Corder JF, Rasmussen S. Ectopia cordis, midline defects and chromosome abnormalities: an epidemiologic perspective. Am J Med Genet 1988;30:811–17.

7. Siliaeva NF, Eye developmental defects in Patau's syndrome (trisomy 13), Oftalmol Zh. 1990;(7):423-6. Russian.

8. Petit P, Fryns JP, Normal/trisomy 13 mosaicism in a 38-year-old male, Genet Couns. 1994;5(3):311-4.

9. Lin HY, Lin SP, Chen YJ, Hung HY, Kao HA, Hsu CH and others, Clinical characteristics and survival of trisomy 18 in a medical center in Taipei, 1988-2004, Am J Med Genet A. 2006 May 1;140(9):945-51.

10. Baty BJ, Jorde LB, Blackburn BL, Carey JC, Natural history of trisomy 18 and trisomy 13: II. Psychomotor development, Am J Med Genet. 1994 Jan 15;49(2):189-94.

11. Peroos S, Forsythe E, Pugh JH, Arthur-Farraj P, Hodes D, Longevity and Patau syndrome: what determines survival?, BMJ Case Rep. 2012 Dec 6;2012. pii: bcr0620114381

12. Cogan DG, Kuwabara T. Ocular pathology of the 13–15 trisomy syndrome. Arch Ophthalmol 1964;72:246–253.

13. Keith CG. The ocular manifestations of trisomy 13–15. Trans Ophthalmol Soc U K 1966;86:435–454.

14. Bocquet L: La pathologie des trisomies D et E. Congr Int Med lang franc. Un Med Can 97:907-927, 1968.

15. Yanoff, M., Frayer, W.C., and Scheie, H.G. (1963). Ocular findings in a patient with 13-15 trisomy. Arch. Ophthal., 70, 372.

16. Smith, D. W. (1963). The No. 18 and Dl trisomy syndromes. Pediat. Clin. N. Amer., 10, 389.

17. Cogan, D.G.,and Kuwabara, T.(1964). Ocular pathology of the 13-15 trisomy syndrome. Arch.Ophthal.,72,246.

18. Keith CG. The ocular manifestations of trisomy 13–15. Trans Ophthalmol Soc U K 1966;86:435–454.

19. Magni R, Pierro L, Brancato R. Microphthalmos with colobomatous orbital cyst in trisomy 13. Ophthalmic Paedi-atr Genet 1991;12:39–42.

20. Kanigowska K, Grałek M, Seroczyńska M, Clinical ocular manifestation of Patau's syndrome (trisomy 13)--own observations, Klin Oczna. 2011;113(7-9):263-5. Polish.

Source of Support: Nil. **Conflict of Interest:** None Declared.

Cite this article as: Aisha Mohammed Alemam, Abdulrahman Alturki, Abdulqader AlHebshi, Sami Alloqmani. Patau Syndrome Survival 6 Years & It's Associated Ocular Findings: Case Report. Int J Med Res Prof. 2018 Jan; 4(1):306-09.
DOI:10.21276/ijmrp.2018.4.1.063