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Case Report

Does NICU Intervention Improve Survivability in Consanguineous Trisomy 13?

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Introduction

Patau Syndrome is a fatal autosomal trisomy ,usually seen because of Meiotic nondisjunction due to maternal advanced age. It is observed at a higher frequency in non-consanguineous union. [1]

Trisomy 13 has a prevalence of 1:12,000–1: 29,000 in newborns.[1,2] Also known as Patau Syndrome, it is a rare and lethal autosomal trisomy 13 with a survivability of only 7-10 days.[3] Only an estimated 9% to 14% of live births survive beyond year 1 of life and are associated with mosaicism.[4,5] Severity of associated malformations also plays a key role in prognosis and survival.[6] It presents with a wide array of dysmorphic features including microphthalmia, cutis aplasia, polydactyly, cleft lip, cleft palate, various congenital heart disease, omphalocele, holoprosencephaly and urogenital abnormalities.[7,8]

There is an intense discussion as to whether timely NICU interventions do play a role in improving mortality in the neonate and long term better prognosis and survivability as a result , especially in VLBW(Very Low Birth Weight) infants.[9-12] There is a higher reporting frequency of Trisomy's in non-consanguineous marriages one example of which is Down Syndrome.[13] Currently diagnosis is based on increasing maternal age, sonographic findings, serum markers and amniocentesis followed in many cases by pregnancy termination as in the West.[14] Risk increases with increasing maternal age with an average of age of 31 years.[15]

Case Report

A 3 days old girl, weighing 3.2 kilograms, was born at term, via C-section ,to a 38 years old mother with a prior history of hypertension, product of a First degree consanguineous marriage, with a birth history of delayed cry, 2 cm Omphalocele, cleft lip, cleft palate, polydactyly-extra digit attached to the little finger in the right extremity and rocker bottom feet(fig.1-3). Patient immediately started de-sating in the low (Sa02) 80s%, became cyanotic, where she was then shifted to the NICU and intubated. Mother denied any drug use or supplements during the pregnancy other than the prescribed antihypertensives.

On examination the patient was tachypneic, had tachycardia, cyanosis and was afebrile. Wide open bulging Anterior Fontanelle, Bilateral intercostal retractions, grunting and extra heart sounds were appreciated. Initial Lab testing revealed a pH of 7.202, pCO2 of 42.7 mmHg, pO2 of 55 mmHg,Hematocrit 55%,Hb 16.5 g/dl, with a TLC of 30.5 cells/ cumm, Platelet count of 373x10^9 per liter (L),Sao2% of 96%,Urea of 21 mmol/L . She was hyponatremic (128 mEq/L), borderline hypokalemic (3.2 mEq/L).

In a couple of days despite intense efforts to resuscitate and maintain the patient in the NICU, condition further decorated with hypokalemia of 2.61 mEq/L, HCO3 11.3 mEq/L, HB 11.5 mg/dl, Thrombocytopenia 56x10^9 per liter (L) and a dropping hematocrit of 33%.

Child was received from the nursery, intubated and started on Ambo bagging. Baby was placed on Ventilator overnight and extubated early morning. Then kept on 02 CPAP. During the course of the day at 11.00 am, severe respiratory distress set in with subcostal retractions and saturation drop to 89% . Intubation was carried out with 3.5mm ETT-oral and tracheal suctioning was done. Patient was placed on the ventilator with PW/CPAP mode and backup ventilator. Post intubation vitals were around HR 160 beats/min, RR 41 breaths/min, spO2 96% and a BSR of 37mg/. Later Pediatric surgical consult was called for to evaluate the omphalocele, which was wrapped with silastic silo and painted. Patient was ordered to be sent to the Pediatric Surgical Ward, with orders for chest and Abdominal X Ray.The neonate continued to deteriorate, became critical and died a couple of days later despite efforts to resuscitate.

Discussion

It is an established fact that advancing maternal age results in increased risk of Meiotic Non Dysfunction resulting in autosomal trisomies, Down's and

Edward's Syndrome to name a few.[16] What this case highlights is an interesting phenomenon of reported trisomy in a consanguineous marriage when higher frequency has been reported in a non-consanguineous setting.[13] It also brings into question the need for aggressive intervention to improve prognosis in terms of morbidity and mortality.90% of live births result in death in year 1 of life and median survival still stands at 7-10 days, and those with higher survival is attributed to mosaicism and severity of associated malformations.[6] It was earlier speculated that adequate, timely intervention may improve survivability and life expectancy.[17] But reports later also suggest the contrary and show an overall decrease in life expectancy.[6,18,19]

No such report has come across taking into consideration NICU interventions and prognosis in Patau Syndrome in Pakistan. From Japan a study revealed better prognosis with NICU interventions.[20] This report impresses on the dire need to improve NICU facilities and study of prognosis and feasibility of NICU intervention in Patau Syndrome.No measures were instituted such as genetic counselling and screening, which further compounded the management in terms of preparedness of the parents.[21]

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