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Prenatal diagnosis of Fraser syndrome: A matter of life or death?

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A female infant was born by natural birth with 46, XX. She was characterized phenotypically by cryptophthalmos, syndactyly, bilateral microtia and ambiguous genitalia. A prenatal ultrasound didn't reveal or raised any suspects for the Fraser Syndrome. It only discovered a unilateral kidney agenesis. At birth the infant showed a severe respiratory distress, intubation was attempted but it failed. The baby was transferred to Santobono-Pausilipon III level hospital. A tracheostomy was performed successfully and saved her life. Computerized Tomography revealed left microphthalmos and a malformation like-coloboma into right ocular globe with cysts and a small calcification parietal anterior. Genetic test revealed the typical mutations in the gene FREM2 confirming the diagnosis of Fraser Syndrome. In her fourth month, after birth, the infant was subjected to an operation to reconstruct eyelids with a mucous membrane graft. The left renal function was normal. The baby showed a delay in motor milestones for visual impairment. At the 19th month follow-up, during a magnetic resonance it was revealed: a normal morphologic brain development, a thin presence in the right optic nerve and the visual cortex were developing.The prenatal diagnosis of Fraser Syndrome is frequently possible. The prenatal ultrasound can reveal features like polyhydramnios or oligohydramnios, echogenic lungs, renal abnormalities or agenesis and cryptophthalmos that are pathognomonic of the Fraser Syndrome. The health providers must keep in mind that if there are suspects of the Fraser Syndrome during prenatal exams, the infants could have a severe malformation in the respiratory tract.

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Human albumin infusions in neonates with gastroschisis in a tertiary government hospital: practices and outcomes

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Objectives: The varying conclusions regarding the utility of albumin in improving outcomes have precluded the consistent use of albumin infusions in gastroschisis. This study aimed to determine the association of albumin infusion and mortality among neonates with gastroschisis admitted in a tertiary government hospital.

Methodology: This was a retrospective cohort study on neonates with gastroschisis admitted from January 2009 to December 2013. A total of 39 records were reviewed and baseline characteristics were described. An association between albumin infusion status and each of the following outcomes were described: length of hospital stay, incidence of at least one complication and mortality.

Results: The results are comparable to earlier studies in that majority had low birth weights, were early term births to young mothers. Most underwent two-stage repair and had hypoalbuminemia. Most (59%) had albumin infusions given postoperatively. Significant differences were noted between neonates given albumin and those who were not given albumin infusions in terms of hospital stay (median Pearson Chi p value 0.027) and the incidence of at least one complication (crude odds ratio 13.2, 95%CI 1.25 – 633.87). There is no significant difference in terms of mortality (crude odds ratio 0.24, 95%CI 0.005 – 2.58).

Conclusion: Human albumin infusion was significantly associated with increased length of hospital stay and higher incidence of at least 1 complication, but with no significant decrease in mortality. However, the small number of data limits further analysis for confounders and modifiers. Larger prospective studies are recommended to further describe these associations.

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