

Tuberous Sclerosis in Pregnancy

A case report and Comprehensive Literature Review

S Mahadasu and S Alomari



Objective

To present a case of pregnancy with tubular sclerosis complex TCS, emphasising the clinical challenges & outcomes and to provide a brief review of the literature on maternal and foetal implications of TSC during pregnancy.

Methods

We report a case of 22 year old primigravida with a known diagnosis of Tuberous sclerosis complex , at the point of diagnosis she had cardiac rhabdomyoma ,epilepsy ,cerebral calcification and confirmed autism ; the pregnancy was planned. A multidisciplinary team including obstetricians, urologist and cardiologist oversaw her care. Maternal and foetal outcomes were monitored throughout pregnancy through clinical evaluations, imaging studies and lab investigations. A review of literature was conducted to conceptualise this case's management and evaluate current recommendations .

Results

At booking , the patient was 5 years free from seizure & AED therapy was discontinued 2 years ago .Anomaly scan showed short long bones and dysgenesis of corpus callosum . At 22 weeks ,aminocentesis and SNP array confirmed the absence of genetic imbalance no copies of the TSC mutated genes were found. Repeated renal scan did not show renal tuber lesions .At 34 weeks of gestation she developed significant proteinuria with a PCR of 34 and mild to moderate pregnancy induced hypertension controlled by labetalol, her PLGF remained normal at 22 , there was an element of mild renal function impairment .The fetal growth was always between 3rd -10th intergrowth centile with normal liquor volume and normal Doppler of the umbilical artery ,at 38+3 weeks labour was induced as she developed symptoms of preeclampsia in the form of headache blurred vision epigastric pain, knee & ankle reflexes were increased, and her blood pressure was increased to 148 / 100 & further deterioration in renal function and she gave vaginal birth to a living growth restricted newborn birth weight at 0.35 intergrowth centile

Discussion

The literature highlights the increased risks of maternal complications such as preeclampsia and seizures, potential foetal fetal inheritance of TSC mutation. increased risk of disease progression , new disease related manifestation and obstetrical complications have also been described in pregnant with TSC Including haemorrhage from ruptured renal tumours ,pre term deliveries pre eclampsia ,preterm premature rupture of membranes ,acute renal failure and placental abruption. In the past various therapeutic approaches were adopted to improve the clinical effects of this disease ,while all of these therapeutic strategies are useful and are still used and indicated ,they are symptomatic therapies based on the individual symptoms of the disease and therefore not fully effective in modifying long term outcomes , however, the introduction of the inhibitors of mTORC1 which allow restoration of the metabolic homeostasis in mutant cells and potentially eliminating most of the clinical manifestation associated with Tuberous sclerosis complex marks a paradigm shift in the management of this disease .Two mTORi ; Everolimus & Sirolimus , FDA class C , lack of adequately controlled studies prevent an accurate assessment of the risk posed to the developing foetus

Conclusion

This case and literature review underscores the importance of individualised care and a multidisciplinary approach in managing pregnancies complicated by TSC. Awareness of potential maternal and foetal risk allows for early intervention and improved outcome. Further studies are needed to establish standardised protocol for the management of pregnancy in TSC.

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Correspondance saboo.h.al-omari@nhs.net