

Successful Pregnancy Outcome in Alagille Syndrome

Raodeo A, Kay T

Dept of Obstetrics and Gynaecology, Royal Devon University Healthcare

Introduction

- Alagille syndrome(ALGS) is a rare autosomal dominant/de novo condition due to mutation in JAG1 or NOTCH2.
- It a multi-system disorder associated with hepatic and renal dysfunction, cholestasis, vertebral anomalies, hypertension, abnormal facial features, cardiac malformations and ocular conditions.
- There is an increased association with miscarriage, pre-eclampsia, obstetric cholestasis and intrauterine fetal growth restrictions.
- There are few documented successful pregnancy outcomes in women with Alagille syndrome

Background

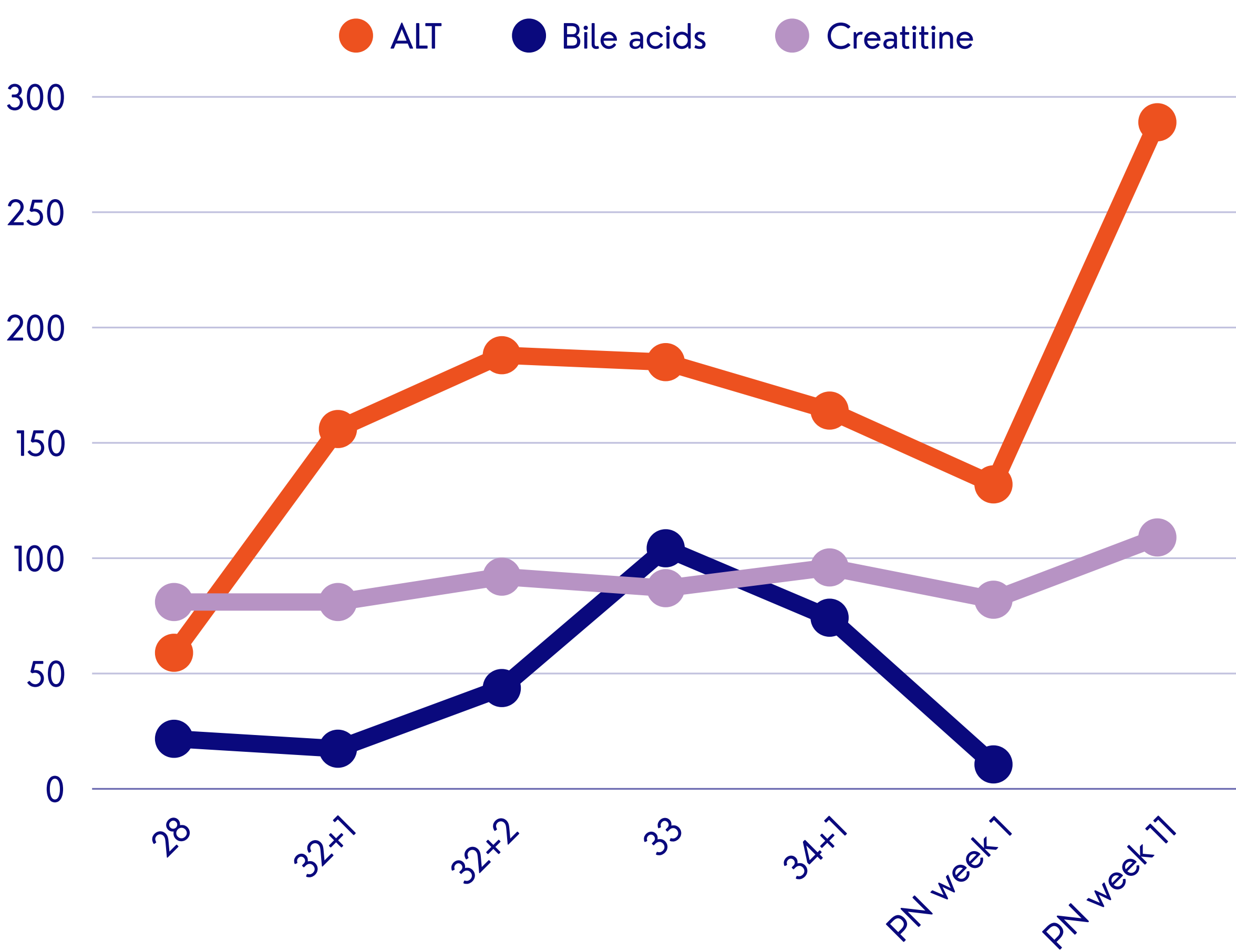
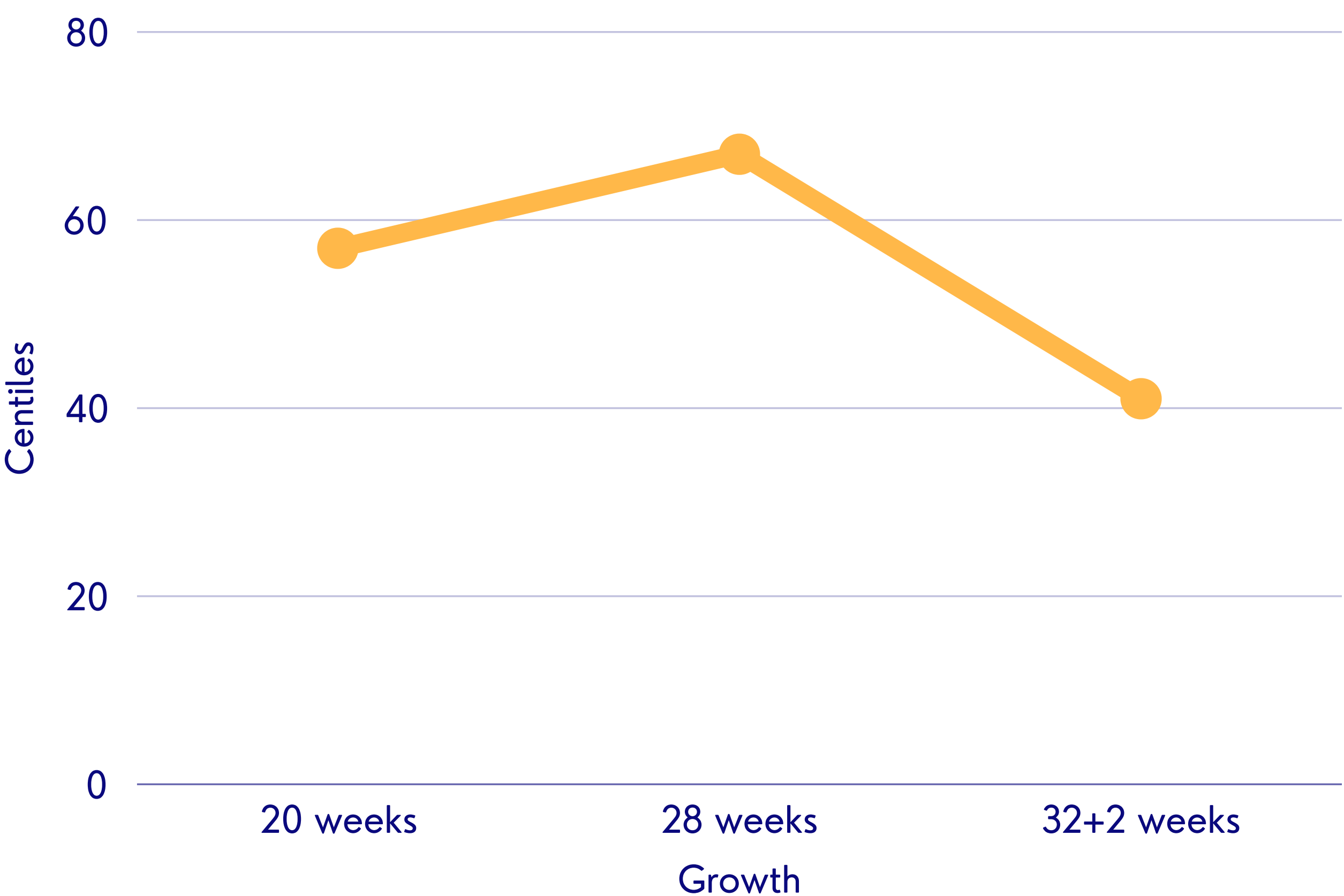
22 years old female-known ALGS manifesting with mild left pulmonary artery stenosis, posterior embryotoxon, low lying cerebellar tonsils, nasal polyposis and some renal & liver involvement.

Referred for consultant led care early-referred to local maternal medicine network. Plan for cardio(advised ECHO-no further concerns) & neuro MDT, with monthly LFTs, FGR pathway & thromboprophylaxis.

First trimester screening was low risk (QUAD test). Anomaly scan normal and uncomplicated first trimester. Regular follow ups with midwife.

Antenatal

- LFTs and bile acid worsening from 32 weeks-admitted for further review
- Anaesthetic input during stay advised not safe for neuraxial procedure.
- Bile acids > 100 with rising urate and proteinuria-planned for induction at 35 weeks
- Growth static on scans
- Spontaneous rupture of membranes at 34+2 with thin meconium hence decision to augment labour



Intrapartum

- On labour ward: Episode of loss of consciousness. Hypertensive and hyperreflexia
- Responding to painful stimuli-suspicion of eclamptic seizure (atypical)
- 2 cm dilated on vaginal exam
- Plan for emergency Caesarean section(C/S) followed by neuroimaging.
- Uneventful recovery with no recollection of events.

Discussion

Despite it's rarity, it's worth considering in women with persistently deranged liver/kidney function postnatally. Furthermore, considerations should be made in creating pathways for pregnant people with ALGS.

Related literature

Morton A, Kumar S. Alagille syndrome and pregnancy. Obstet Med. 2021 Mar;14(1):39-41. doi: 10.1177/1753495X19849738. Epub 2019 Jun 9. PMID: 33995572; PMCID: PMC8107961.