Anaesthesia in an adult with Rubinstein–Taybi syndrome

Editor—Twigg and Cook have made an interesting contribution to anaesthesia for Rubinstein–Taybi syndrome in an adult. This complements reports of anaesthesia in children with this syndrome. Baer and colleagues described anaesthesia in two children and reported no problems. Airway problems may be expected, as Hennekam and colleagues found micrognathia or retrognathia in about half of the 45 patients they reviewed. A further paper by these workers showed 84% of these patients have microstomia, which might have an effect on laryngoscopy. These two reports are in children and the descriptions of anaesthesia are sketchy. Hennekam and colleagues, in a series of 45 cases, recorded three instances of difficulties attributable to laryngeal collapse and difficulty in intubation. Stevens and colleagues reviewed the medical problems of 50 such patients and noted frequent hospitalizations and operations (210), showing that although Rubinstein–Taybi syndrome is rare, each patient may undergo multiple anaesthetics, an average of 2.7 each. He reported problems in nine cases. These included respiratory distress, apnoea and prolonged anaesthesia.

Hennekam’s team has also elucidated the genetic deficit. This is a microdeletion on chromosome 16. Recent interest in the human genome project means that this segment of chromosome has now been sequenced. The protein from the affected gene is CREB binding protein. This protein is used in the biochemistry of every cell in that it assists cAMP and is needed to read DNA. The mutation halves the amount present in each cell. This is termed haplo-insufficiency. It is remarkable that disruption of this protein at a fundamental point in the cell cycle gives a syndrome as benign as Rubinstein–Taybi. Hennekam found that the incidence of the disease was around 1 in 90,000 (personal communication to ORD).

Twigg discusses Stirt’s report on cardiac arrest after the use of succinylcholine in these children. However, in another of their references, Critchley and colleagues used it without ill effect. So the jury is still out on the question of the safety of succinylcholine in these patients.

We had previously found a laryngeal mask very suitable in a patient with Rubinstein–Taybi syndrome. Our patient had a slow recovery after we gave an anaesthetic with propofol and spontaneous respiration. We notice that Twigg and Cook took precautions against aspiration. They used an H2 blocker as a premedicant and passed an orogastric tube; but when their patient was found to be a grade 2 view at direct laryngoscopy and so could have been intubated easily, they did not intubate. We wondered how they would have defended their anaesthetic if their patient, whom they had previsaged might vomit and aspirate, had gone on to do so. While we congratulate Twigg and Cook on the successful outcome of this case, we feel that an unnecessary risk was taken in the conduct of the anaesthetic.

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Editor—We are grateful for Dr Dearlove and Dr Perkins’ interest in our report and for the opportunity to reply to their comments. They agree with our findings that there are few reports in the literature and that these relate to paediatric problems, with no reports of anaesthetic techniques used in adults. It is indeed noteworthy that the genetic defect involving the CREB binding protein is at a fundamental part of the cell cycle, and involves chromosomal rearrangements of the cytogenetic band 16p13.3, with an incidence at around 1 in 125,000 births.

The phenotypic syndrome of Rubinstein–Taybi syndrome is markedly variable, and we would suggest that an anaesthetic assessment needs to be made on an individual basis. The patient we described had no history of reflux, aspiration or recurrent chest infections, and we considered her to be at more risk from difficult intubation than regurgitation. However, communication problems meant that an entirely reliable history was not available. The use of an H2 blocker as a premedicant was sensible because the potential risk of regurgitation, however small, could not be eliminated. The passage of an orogastric tube, likewise, was safe and prudent and confirmed that the stomach was empty. The choice of ProSeal LMA (PLMA) was made on the basis of it being the best airway device for this particular operation (bilateral phacoemulsification), in this particular patient. We had no objective evidence to support a history of reflux, but the patient had clear physical findings suggesting a high risk of difficulty with intubation. Had we considered the risk of regurgitation to be high, we would have had to choose between a rapid sequence induction in a patient with a high potential for difficult intubation, or an awake fibreoptic intubation in a patient with marked comprehension difficulties. Laryngoscopy was performed in order to document the degree of difficulty in obtaining a view of the glottis, should the patient require intubation in the future. We believe that all measures were taken to minimize potential and actual risk during this anaesthetic and that the correct technique was chosen.

Rather confusingly, Dearlove and Perkins report using a classic laryngeal mask airway (LMA) in a patient with Rubinstein–Taybi syndrome. Without further details, it is difficult to comment and certainly we would not criticise this, but we are somewhat surprised at their criticism of our use of the PLMA in the face of their use of a classic LMA. The case they report would likely have taken place before the PLMA became available and presumably, like our patient, was judged clinically to have a low risk of regurgitation of gastric contents. In individual cases, with appropriate assessment, it may be that either the classic LMA or PLMA may be suitable for use in patients with Rubinstein–Taybi syndrome. However, the PLMA is a useful addition to the airway armamentarium, and as it is designed to separate the gastrointestinal and respiratory tracts, compared with the classic LMA, it may increase the margin of safety in those patients with Rubinstein–Taybi syndrome in whom use of a tracheal tube is not considered necessary.

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1 LMA is the property of Intavent Limited.
7 Stirn JA. Succinylcholine in Rubinstein–Taybi syndrome. Anesthesiology 1982; 57: 429

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