Cataract in Childhood Glaucoma and Anterior Segment Dysgenesis

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Cataracts can occur in association with ocular or systemic comorbidities. Presence of these comorbidities may alter the presentation and management of lens conditions. In this chapter we will be discussing two such comorbidities: childhood glaucomas and anterior segment dysgenesis. Although childhood glaucoma and anterior segment dysgenesis are separate entities, they have been clubbed together since anterior segment dysgenesis is associated with glaucoma in significant number of cases and few cases of presumed primary congenital glaucoma may actually be glauocomas secondary to anterior segment dysgenesis.

11.1 Childhood Glaucoma

Primary congenital glaucoma (PCG) is an anomaly affecting anterior chamber angle which leads to obstruction of aqueous outflow, increased intraocular pressure (IOP) and optic nerve damage [1]. Buphthalmos is used to describe visible enlargement of the eyeball at birth or early childhood due to an uncontrolled glaucoma [2]. High intraocular pressure (IOP) causes increase in axial length and corneal dimensions of the eye, leading to axial myopia, stretched limbus, corneal thinning, and visibly enlarged eyeballs. The common causes of buphthalmos include primary congenital glaucoma (PCG), Sturge–Weber syndrome, neurofibromatosis and aniridia [2].

The incidence of PCG is one in every 10,000–15,000 live births which accounts for 0.01–0.04% of total blindness [3]. It is bilateral in up to 80% of cases and two-thirds of the cases are males. Most cases are sporadic (90%) [4]. However, in the remaining 10% there appears to be a strong familial pattern.

11.1.1 Clinical Presentation

Vision loss in glaucoma eyes in children may occur secondary to an uncorrected refractive error, corneal opacity, optic nerve damage, amblyopia or per se cataracts. Examination findings may be variable. However, following points should be noted:

- Progressive myopia and high astigmatism may be seen.
- Cornea may be hazy and presence of Haab striae is a common finding (Fig. 11.1), which may obscure vision.
- Cataract may be anterior or posterior subcapsular cataract, total or less commonly cortical or zonular cataract (Fig. 11.2). Cataract may be primary or more commonly secondary to trabeculectomy in 6–58% cases [5–7].
- Bleb in post trabeculectomy eyes with buphthalmos may be thin cystic and rarely associated with other complications (Fig. 11.3).
- Fundus examination may reveal advanced cupping which may be reversible (to some extent) in small children. Features associated with pathological myopia (e.g. tessellated fundus) may be seen.

11.1.2 Investigations

In addition to usual investigations, ultrasound biomicroscopy can aid in measurement of angle to angle and bag diameter to plan for surgery (Fig. 11.4). UBM also helps to assess anterior segment structures, anterior chamber depth (ACD), angle anomalies, abnormal iris insertion, sulcus-to-sulcus measurement and identifies lax zonules and/or pre-exisiting posterior capsular defect preoperatively [8].

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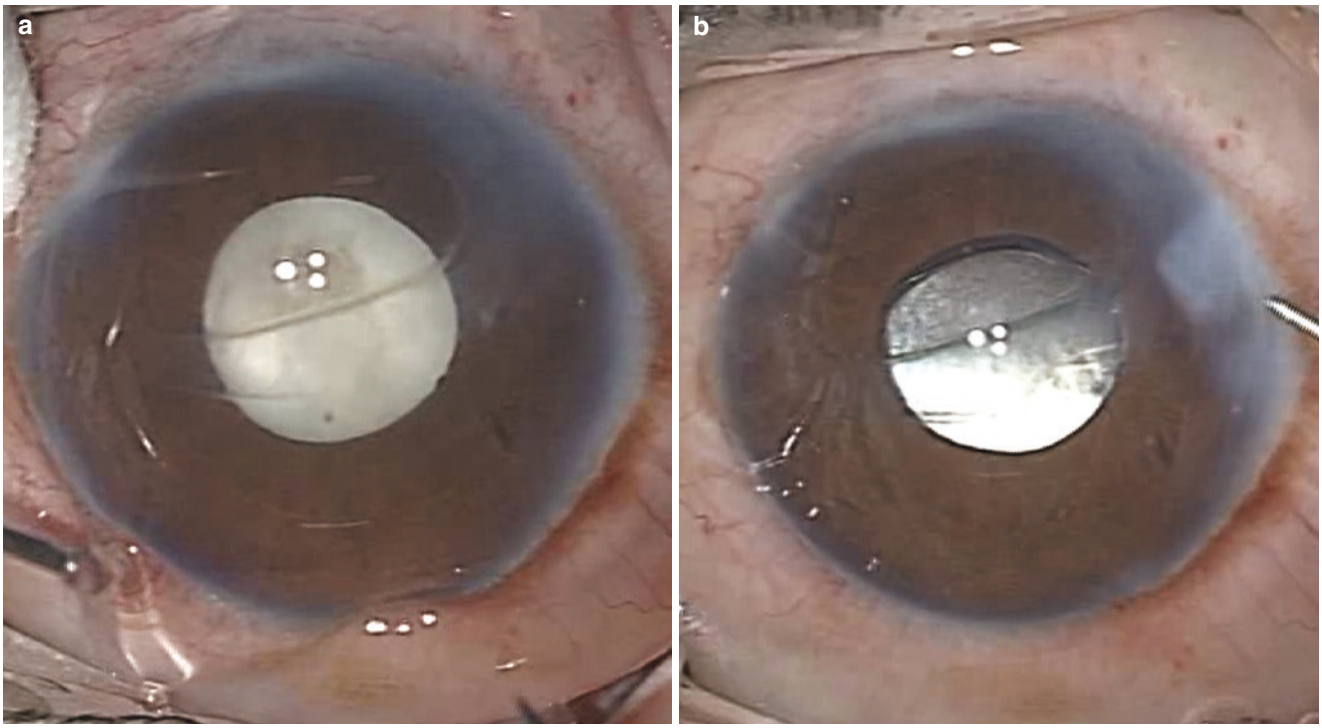

Fig. 11.1 Congenital glaucoma with Haab striae and total cataract. (**a**) Preoperative picture with large superior and multiple peripheral Haab striae seen in cornea, (**b**) postoperative picture of same case with IOL in situ and more pronounced Haab striae

11.1.3 Management of Cataract in Buphthalmic Eyes

The surgical challenges faced by pediatric surgeons while operating for cataract in buphthalmic eyes include the following:

- Corneal haze may cause difficulty in visualization during surgery (Fig. 11.5).
- Very deep anterior chamber (AC) result in difficult instrumentation. Intraoperatively, anterior chamber depth may show frequent fluctuations due to low scleral rigidity in these stretched eyes.
- Phacodonesis, lax lens zonules, liquefied vitreous, and, thus, a weak posterior capsular support can lead to inadvertent complications. There is increased risk of vitreous loss in these patients.
- Highly elastic anterior and posterior capsule may cause continuous curvilinear capsulorhexis (CCC) to be a challenging step.
- Wound closure in eyes with raised pressure may be difficult. All wounds must be well-sutured at the end of surgery in these eyes as postoperatively shallow AC is often noted.
- Intraocular lens (IOL) power calculation remains difficult. Post trabeculectomy buphthalmic eyes have a shift towards with-the-rule astigmatism [9, 10]. As most of these eyes are high myopic, IOL power calculation should be done using appropriate IOL formulae, e.g., SRK-T for axial lengths >24.5 mm. Many a times, no single IOL power formulae might be able to predict the correct emmetropic power of implant for buphthalmic eyes.
- Large eye size and bag dimensions that may lead to postoperative intraocular lens (IOL) decentration [11, 12]. But if bag size appears to be large on UBM, surgeon should plan multipiece IOL in sulcus with optic capture in bag (i.e., optic in bag and haptic in sulcus) (Fig. 11.6). Iris fixated and customized IOLs have also been described for such eyes [12].
- Postoperatively, there are high chances of visual axis opacification (VAO) formation owing to an inflammatory response and formation of dense capsular fibrosis.

11.1.4 Surgical Outcomes

Temporary cessation of ocular growth is reported after adequate IOP control in eyes with AL > 22 mm and in children aged 3 months or older [13].

Our experience with 31 eyes of primary congenital glaucoma (post trabeculectomy) with visually significant cataract undergoing lens aspiration surgery showed a mean best corrected visual acuity of 6/60 (Snellen's) at one year postoperatively. Reasonably predictable refractive results were obtained in these eyes, provided intraocular pressure was well controlled [14].

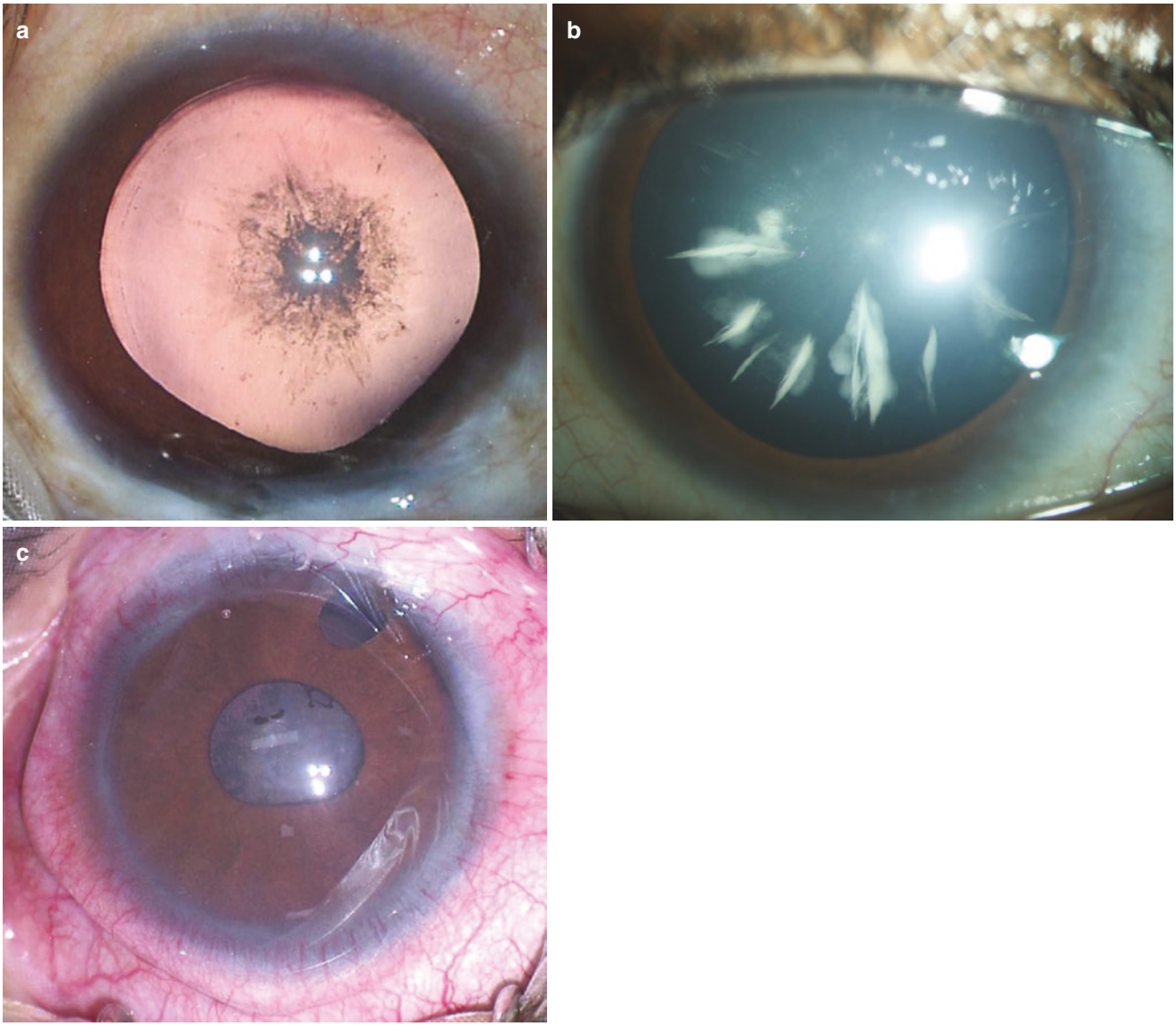

Fig. 11.2 Morphology in cataract with childhood glaucoma post trabeculectomy. (**a**) Posterior subcapsular cataract, (**b**) cortical cataract, (**c**) diffuse cataract with large superonasal peripheral iridotomy

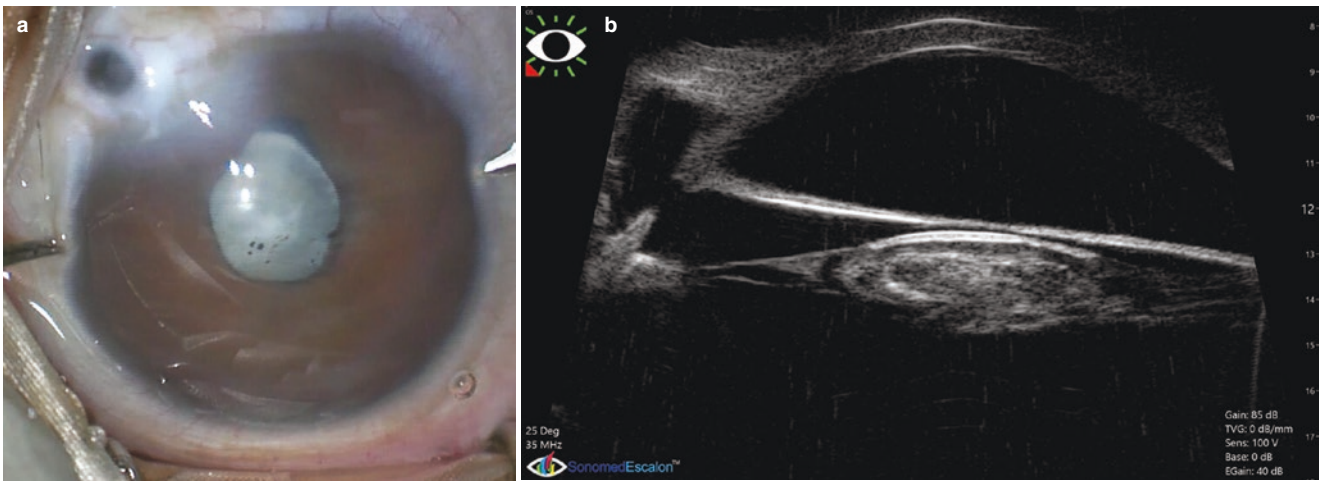

Fig. 11.3 Post trabeculectomy thin cystic bleb with iris prolapse from the ostium and total cataract. (**a**) Clinical picture, (**b**) ultrasound biomicroscopy of the same case showing patent ostium and elevated bleb

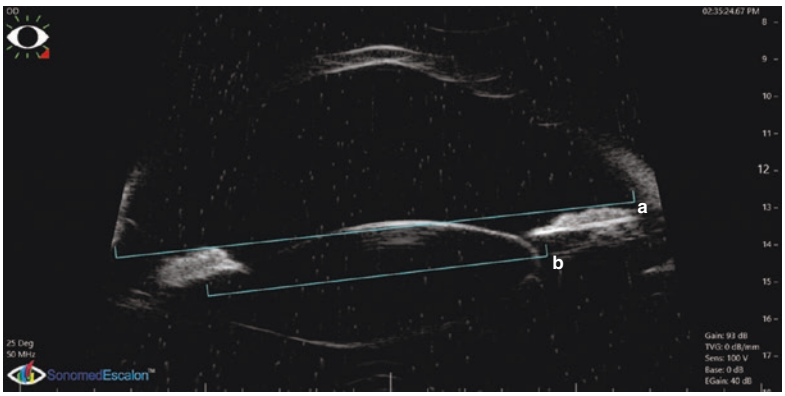

Fig. 11.4 Ultrasound biomicroscopy showing dimensions of a buphthalmic eye. Complete white-to-white examination is not possible in single view. (**a**) angle to angle distance, (**b**) bag diameter

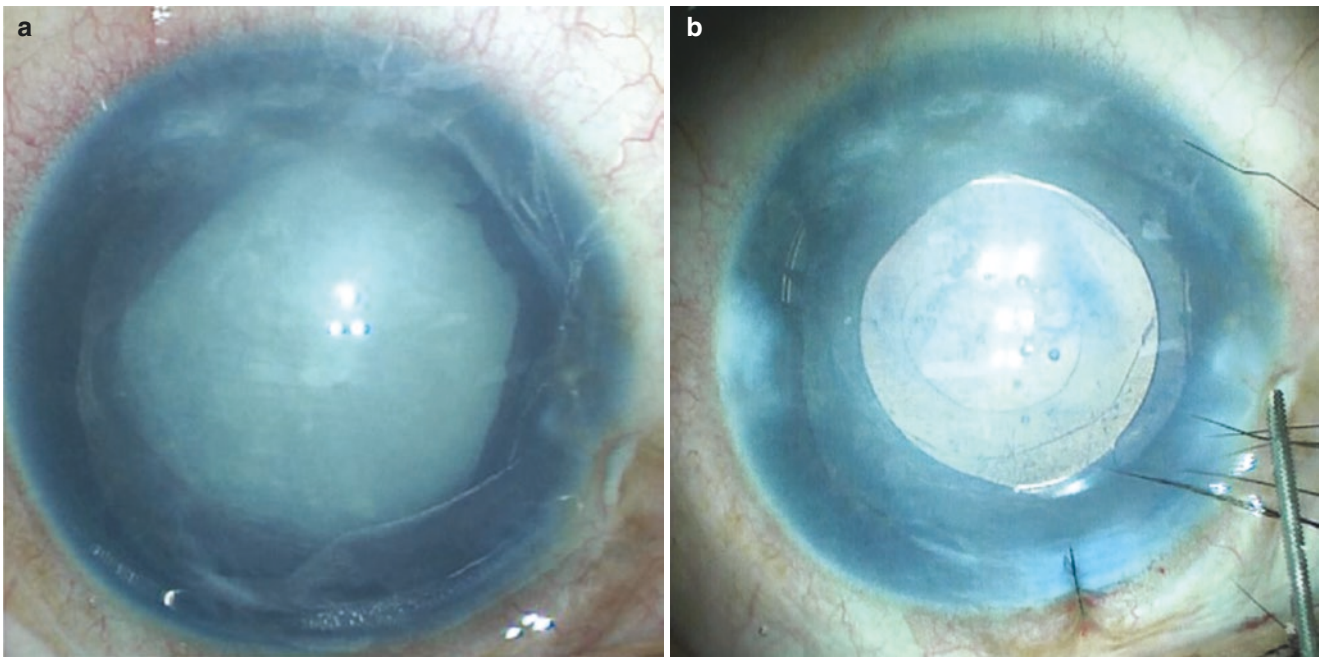

Fig. 11.5 A 6-year-old child with congenital glaucoma with corneal haze with atrophic iris. (**a**) Preoperative picture with total cataract, (**b**) postoperative picture with circular anterior and posterior capsulorhexis and IOL in bag

Thus, besides control of IOP, visual rehabilitation of buphthalmic eyes involves appropriate management for amblyopia, keratoplasty for corneal opacity, and timely cataract surgery for visually significant cataract. Buphthalmic eyes undergoing cataract surgery can achieve successful refractive and visual outcomes if careful preoperative planning is carried out regarding the choice of IOL type and IOL power, taking into consideration the adequacy of intraocular pressure control, accurate biometry, assessment of bag size, and use of appropriate IOL power formulae.

11.2 Anterior Segment Dysgenesis

Anterior segment dysgenesis (ASD) is a group of disorders arising from abnormal development in cornea, iris, lens, and angle structures. This spectrum includes Axenfeld's anomaly, Rieger's anomaly, Axenfeld–Rieger syndrome (ARS), Peters anomaly, sclerocornea, aniridia, posterior keratoconus, and iridogoniodysgenesis. They occur often due to abnormalities in neural crest differentiation and migration. Various classification systems are used for describing ASD depending either on their clinical features or area of involve-

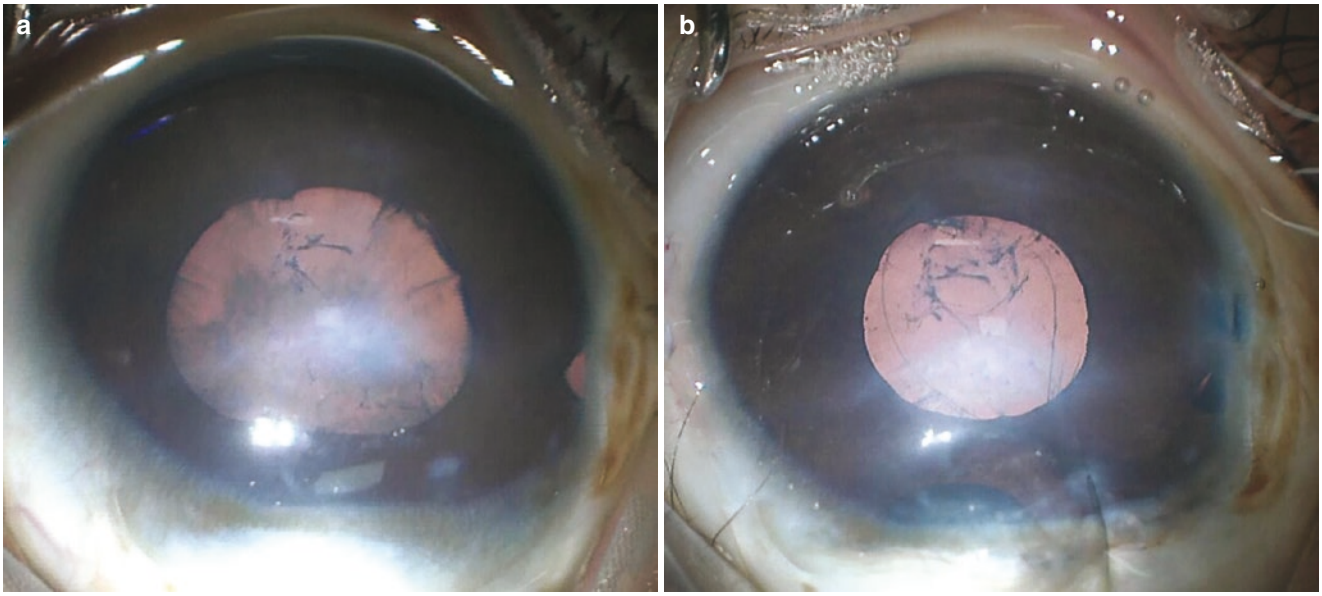

Fig. 11.6 A 15-month-old child with congenital glaucoma with corneal haze with Haab striae and large cornea (buphthalmos). (**a**) Preoperative picture with anterior and posterior subcapsular cataract, (**b**) postoperative picture with IOL in sulcus with optic capture with anterior and posterior capsulorhexis (bag complex) for better centration

ment [15–17]. Lens abnormalities are not uncommon in cases with ASD. Townsend, Font and Zimmerman have classified ASD based on involvement as Descemet layer defect alone or associated with lens abnormalities or with iris stromal abnormalities. This involvement of lens suggested the effects of primary mesenchymal defect on the development of lens [17].

11.2.1 Embryology

The surface ectoderm of developing human embryo invaginates and forms lens vesicle in the embryonic cup at sixth week of gestation. Then, neural-crest derived tissue migrates in three waves beneath this surface ectoderm. The surface ectoderm forms the corneal epithelium. The three waves forms endothelium, corneal stroma, and iris stroma. Any arrest in the development of these layers may affect further development of anterior chamber leading to different presentations of ASD [18].

11.2.2 Genetics

Many genes are involved in the ASD with variable degrees of penetrance. Forty percent cases occur due to involvement of PITX2 (4q25) and FOXC1 (6p25). Typically, PITX2 disruption is associated with ARS with ocular and dental abnormalities, and FOXC1 is associated with ARS with hearing or cardiac abnormalities. Others associated with ARS include PAX6 (11p13) and FOXO1A (13q14) [19, 20]. ARS has autosomal dominant inheritance pattern in 70% cases. In Peter's anomaly, rare cases have been attributed to PITX2, FOXC1, and PAX6 mutations, but the majority of cases are sporadic [21–23].

11.2.3 Clinical Features

- Axenfeld–Rieger syndrome.

Axenfeld anomaly presents as posterior embryotoxon (Fig. 11.7) (anteriorly displaced Schwalbe's line) and iris strands adhered to the anteriorly displaced Schwalbe's line. Rieger anomaly includes posterior embryotoxon, pseudopolycoria, and iris atrophy (Fig. 11.8) while Rieger syndrome is Rieger anomaly along with systemic findings including facial bone defects, hypertelorism, telecanthus, maxillary hypoplasia, dental abnormalities (microdontia and hypodontia), umbilical abnormalities, or pituitary involvement. Thus, they are now considered as a spectrum of disorder termed as Axenfeld–Rieger syndrome (Fig. 11.9). It may vary from subtle changes in the angle to severe ocular changes. Systemic involvement may also include cardiac and endocrine system. Fifty percent cases with ARS are associated with glaucoma [24, 25].

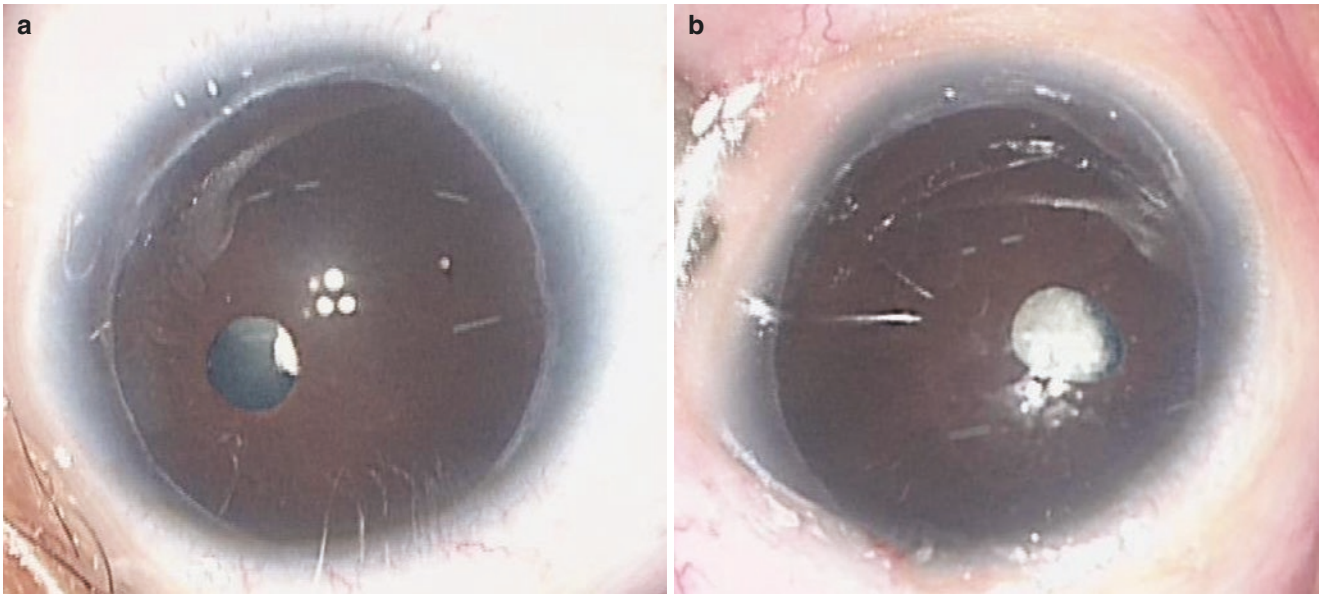

Fig. 11.7 Mild variant of Axenfeld–Rieger syndrome. (**a**, **b**) Posterior embryotoxon in a 9-month-old child with cataract in right and left eye, respectively. Also notice presence of corectopia in both eyes

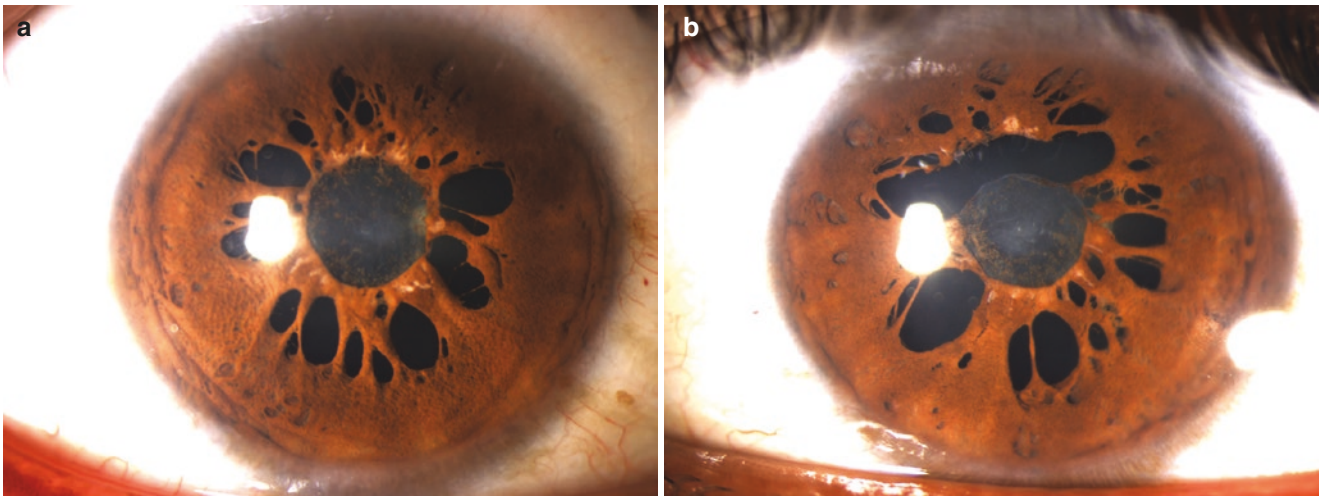

Fig. 11.8 (**a**, **b**) Rieger anomaly—polycoria and iris atrophy in a 7-year-old girl in right and left eye, respectively

- Peters Anomaly.

Peter syndrome is characterized by a shallow anterior chamber, synechiae between iris and cornea, and central corneal opacity. It occurs due to defect in endothelium, Descemet membrane and posterior stroma due to the defect in the migration of the neural crest cells. This syndrome can vary in severity with ocular findings ranging from unilateral mild central corneal opacity to severe bilateral microphthalmia, corneal opacification, cataract, and glaucoma. Eighty percent cases have bilateral presentation. The Peters anomaly has been further divided into type I and type II. Type I Peters anomaly is categorized by central corneal opacity and iridocorneal adhesions (Fig. 11.10). Type II Peters anomaly has a more severe phenotype with corneal opacity and lens involvement with iridocorneal touch with or without cataract (Fig. 11.11). The Peters plus syndrome includes the anterior segment findings with systemic developmental anomalies. These include craniofacial dysmorphism, cleft lip/palate, short stature, brachydactyly, ear abnormalities, and mental retardation [26, 27].

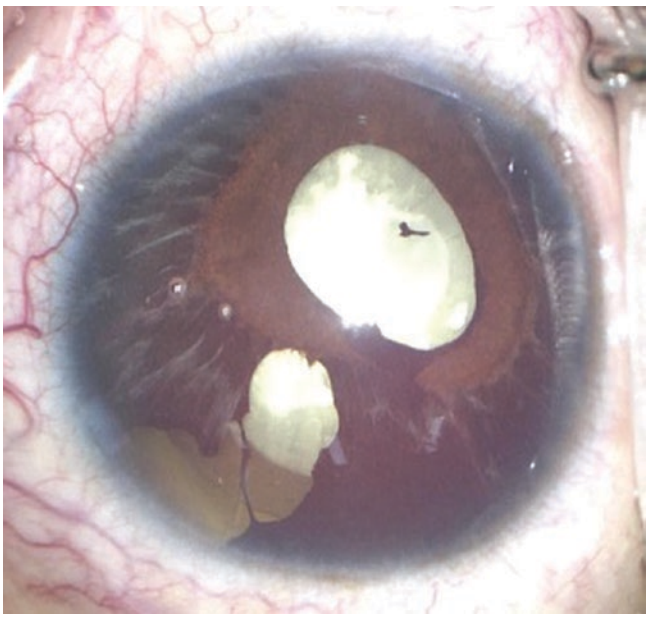

Fig. 11.9 Severe variant of Axenfeld–Rieger syndrome—posterior embryotoxon with corectopia, iris atrophy and polycoria along with total cataract

- Aniridia.

Aniridia is a rare congenital disorder characterized by iris hypoplasia along with other abnormalities of the eye [28] . Ocular abnormalities include dry eye, aniridia associated keratopathy (AAK) (Fig. 11.12), angle abnormalities, glaucoma, cataract, foveal hypoplasia, optic nerve hypoplasia, nystagmus, or strabismus [29–31]. Cataract morphology may be anterior or posterior subcapsular, lamellar, cortical, total or a combination of the above [28] (Fig. 11.13). Zonular weakness may be seen and ectopia lentis may be associated in some patients [28]. This can be managed with placement of capsular tension ring in mild cases (Fig. 11.14). Anterior polar or pyramidal cataract may be associated with aniridia along with remnants of persistent fetal vasculature [32] (Fig. 11.15).

11.2.4 Differential Diagnosis

The differential diagnosis of ASD includes obstetric trauma, congenital glaucoma, intrauterine infections like rubella, herpes simplex virus, and bacterial infections, iridocorneo-

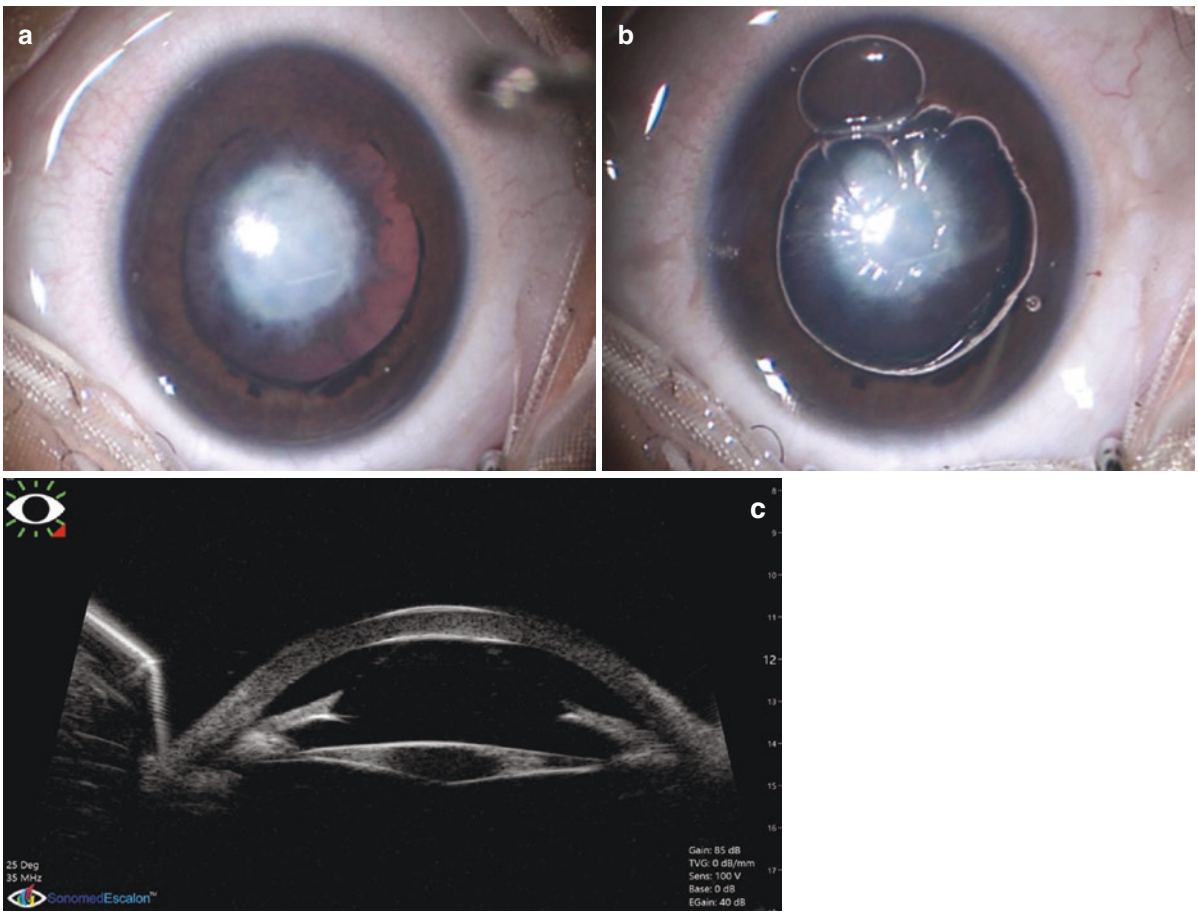

Fig. 11.10 Peter's anomaly type 1 in 2-month-old child. (**a**) Small corneal opacity with iridocorneal adhesions with cataract. (**b**) Intraoperative picture after injection of air in anterior chamber, irregular air bubble is seen due to iridocorneal adhesions. (**c**) Ultrasound biomicroscopy of the same showing fine central iridocorneal adhesions

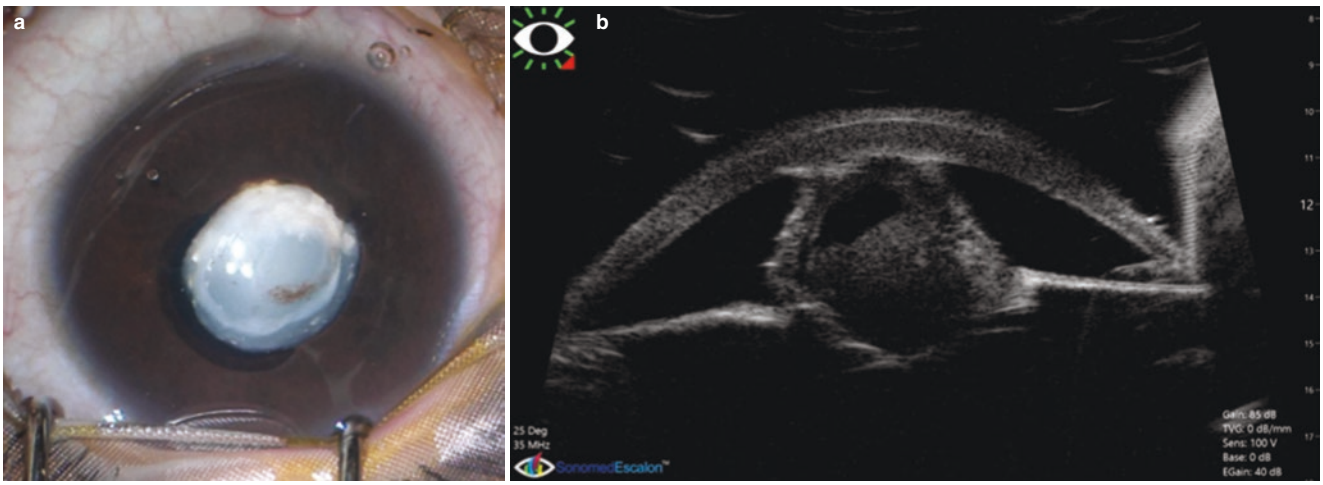

Fig. 11.11 Peter's anomaly type 2 in 4-month-old child. (**a**) Central corneal opacity with total cataract, (**b**) ultrasound biomicroscopy of the same showing iridolenticular adhesions with anterior displacement of lens

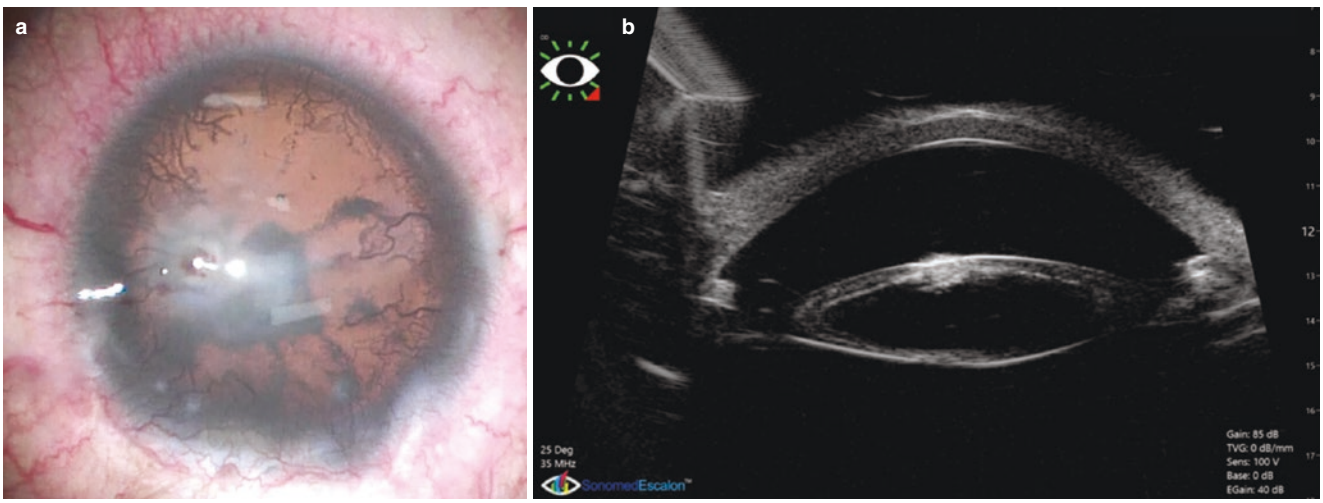

Fig. 11.12 Aniridia associated keratopathy with corneal opacity with 360° pannus. (**a**) Clinical picture, (**b**) ultrasound biomicroscopy of the same showing anterior subcapsular cataract, not clearly seen clinically

endothelial syndrome (Fig. 11.16), metabolic diseases like mucopolysaccharidosis, mucolipidoses, and tyrosinosis, congenital hereditary endothelial dystrophy, congenital hereditary stromal dystrophy, and dermoids.

11.2.5 Investigations

Apart from usual investigations ultrasound biomicroscopy may allow us to preoperatively assess the area beneath the corneal opacity. It helps us to determine the area of corneal opacity, depth of opacity, presence of iris adhesion, anterior chamber depth and angle details in the involved area. The lens can be visualized and observed for kerato-lenticular adhesion or presence of any tilting of the lens (Figs. 11.10b, 11.11b, and 11.13d, e). This can help us in the planning of the cataract surgery and expect a better outcome.

11.2.6 Surgical Pearls

Patients with ASD should be screened for glaucoma and managed appropriately. They require optimization of visual function which includes refractive error prescription and tinted contact lenses for photophobia. This is important for

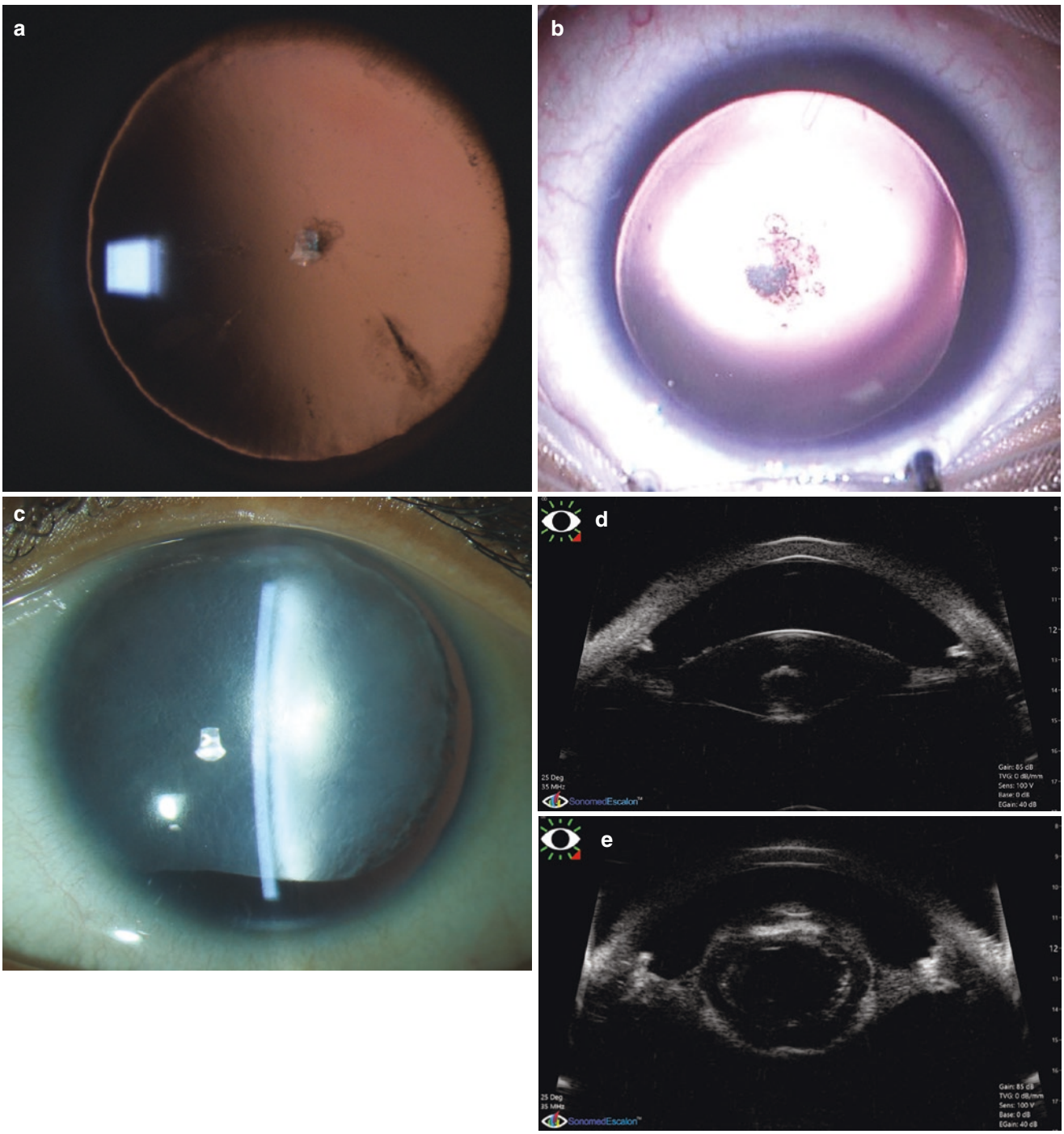

Fig. 11.13 Aniridia with cataract. (**a**) Clinical picture of insignificant anterior polar with cortical cataract. (**b**) Clinical picture of posterior subcapsular cataract. (**c**) Clinical picture of total cataract with inferior notching due to zonular laxity (Pseudo-lens coloboma). (**d**) UBM of aniridia patient showing minimal cataract and remnant of iris stump clearly with no subluxation. (**e**) UBM of aniridia patient with anterior polar and zonular cataract with zonular laxity causing increase in lens globularity

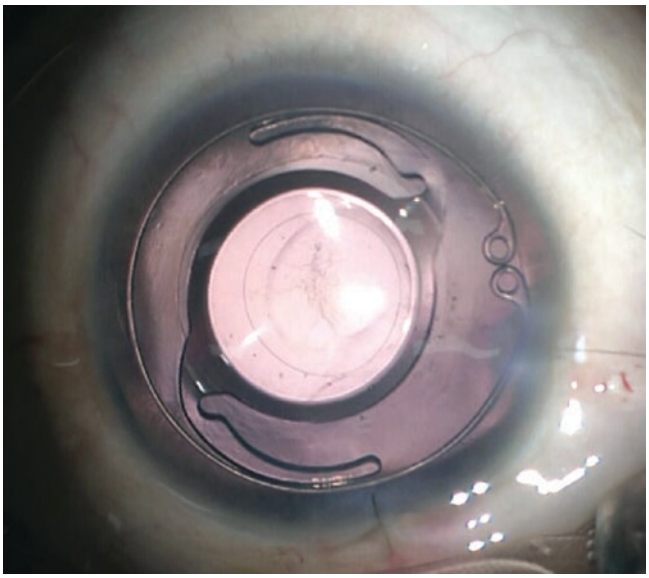

Fig. 11.14 Postoperative picture of aniridia with mild subluxation. Notice anterior and posterior capsulorhexis with well-centered IOL in bag with capsular tension ring

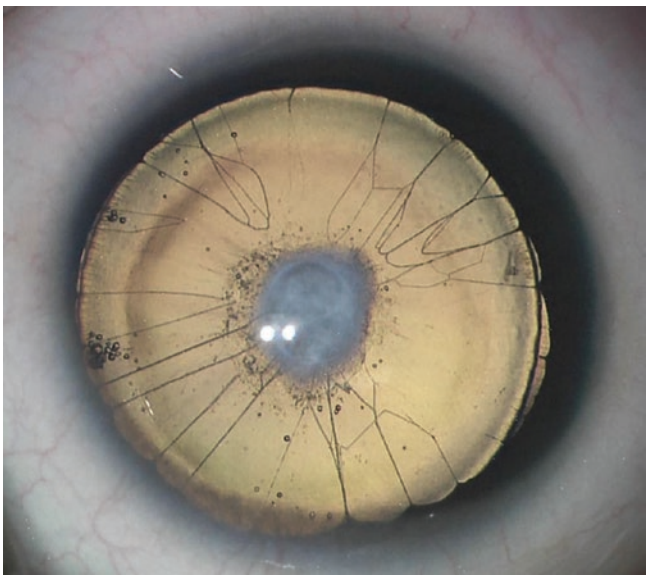

Fig. 11.15 Aniridia with anterior polar cataract with remnant of persistent fetal vasculature

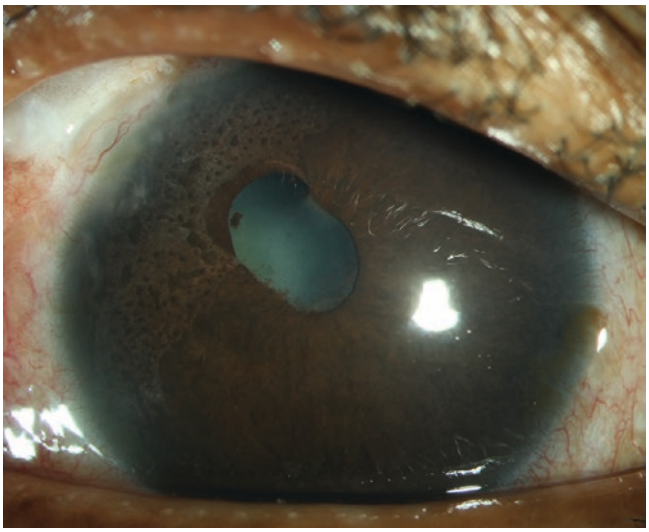

Fig. 11.16 A 16-year-old girl with Cogan Reese syndrome. Notice atrophic iris and corectopia with iris nodules

prevention and treatment of amblyopia. Few patients may also require surgery for corneal opacity, lens abnormality, or glaucoma management. Various challenges may be involved in the cataract surgery in cases with ASD.

- Corneal abnormalities: corneal opacity or aniridia associated keratopathy (AAK) may cause difficulty in anterior chamber visualization. Staining of the anterior capsule enhances its visualization during capsulorhexis. Other methods like use of illumination techniques like transcorneal oblique illumination or endoscope-assisted surgery can help in better visualization but are time-consuming methods with a greater learning curve [33–35]. Image-guided surgery using femtosecond laser for cataract surgery in Peter's syndrome has also been recently used [22]. However, the depth and height of the femtosecond laser should be cautiously adjusted to avoid damage to the endothelium.
- Presence of kerato-lenticular adhesion increases difficulty in surgical maneuvering. There may be risk of Descemet and endothelial damage during release of the keratolenticular adhesions and difficulty to achieve appropriate size regular capsulorhexis. We have also

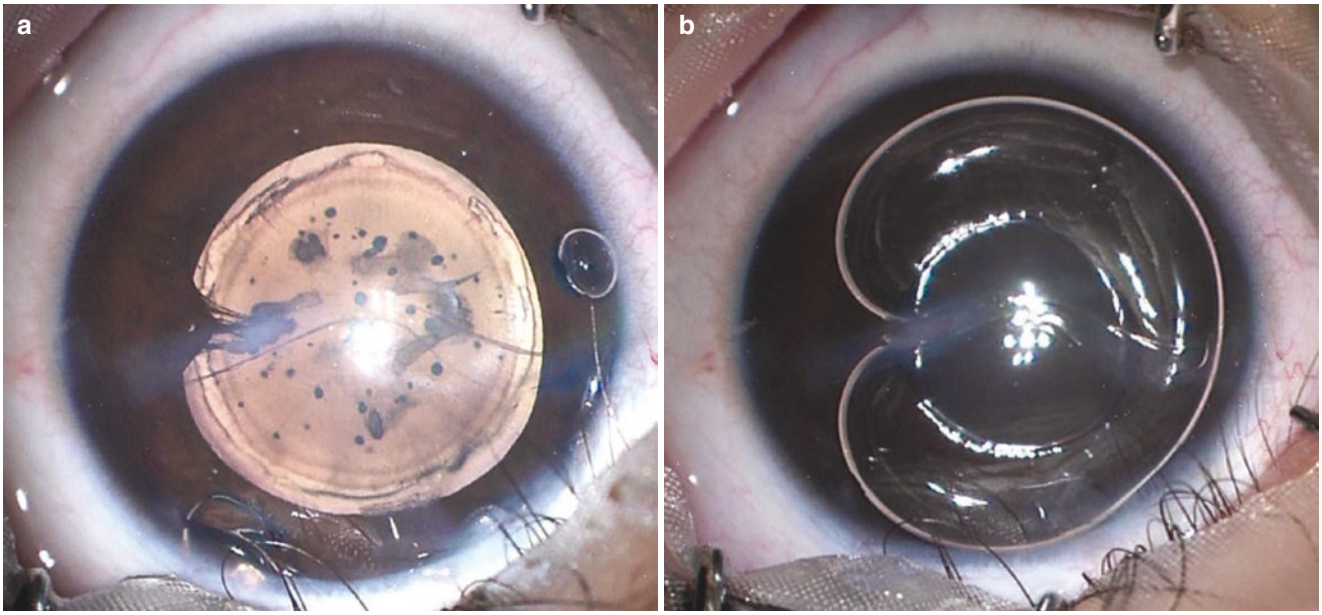

Fig. 11.17 Irregular air bubble sign in anterior segment dysgenesis with glaucoma. (**a**) Corneal opacity with iridocorneal adhesions with Haab striae with anterior capsular pigments and zonular cataract. (**b**) Intraoperative picture after ingestion of air in anterior chamber, irregular air bubble is seen. Iridocorneal adhesions which were not clearly seen preoperatively are enhanced

noticed "irregular air bubble" in anterior chamber as a sign of presence of adhesions when they are not clearly visible (Fig. 11.17a, b).

- Iris abnormalities like corectopia, polycoria, iridocorneal adhesion, posterior synechiae between iris and lens may require anterior segment reconstruction together with synechiae release, anterior chamber formation or pupilloplasty during cataract surgery. Aniridia patients require use of tinted glasses or contact lenses postoperatively. Iris prosthetic devices may be used [36]. There is risk of secondary glaucoma, corneal decompensation, band-shaped keratopathy and device displacement [37] (Fig. 11.18).
- Glaucoma management in cases with ASD is of importance and may include medical management, surgical management or both. Thus, a regular follow-up with monitoring of visual acuity and the intraocular pressure is crucial.

Challenges in surgery in patients with ASD have to be carefully dealt with in order to achieve satisfactory visual outcomes. In addition to cataract surgery, glaucoma management is of utmost importance in these cases.

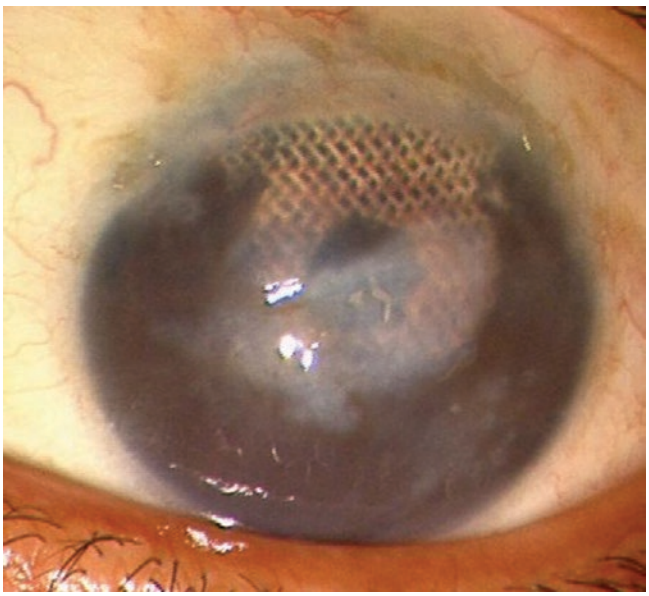

Fig. 11.18 One year postoperative picture of patient operated with iris implant (outside center) with acquired aniridia (traumatic) with band-shaped keratopathy and corneal decompensation

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