

[1] MARFAN SYNDROME

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Abstract not available at the time of print.

[2] VON HIPPEL-LINDAU DISEASE: DIAGNOSIS AND CLINICAL MANAGEMENT

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von Hippel-Lindau disease (vHL) is a hereditary tumor predisposition caused by mutations in the *VHL* tumor suppressor gene. The most common manifestations are benign hemangioblastomas in the cerebellum, spinal cord, and retina. Patients are also at risk of renal cell carcinoma, pheochromocytoma, pancreatic neuroendocrine tumors, and endolymphatic sac tumors of the inner ear. As many as 70% develop their first manifestation in childhood, most often a retinal hemangioblastoma, which may be the only sign of the disease for years. The ophthalmologist will often be the first doctor to meet a potential vHL patient, and plays an important role in assessing the patient's likelihood of having vHL. The disease is important to recognize early, as the mainstay of vHL management is prophylactic surveillance of the patient and predisposed family members.

[3] NEUROFIBROMATOSIS AND OPTIC GLIOMAS

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Optic pathway gliomas can occur in children with and without neurofibromatosis type 1. The screening of NF1 patients and the management of OPGs have been challenges facing paediatric ophthalmologist working with their oncology colleagues. LGG2 (Low Grade Glioma SIOP Trial 2) ran between 2004 and 2013. Prospectively collected ophthalmic data has helped to offer a better understanding of the natural history of these tumours and a more informed approach to parental counselling.

[4] NF1-ASSOCIATED OPTIC GLIOMAS, TREATMENT STRATEGIES

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15-20% of children with NF-1 develop optic pathway gliomas (OPG), of which 13-15 % are treated with chemotherapy. Overall survival is excellent, but a significant proportion of both treated and untreated patients have compromised visual outcome.

A main aim of chemotherapy has been to improve visual outcome. However, data is sparse concerning the efficacy of chemotherapy for this indication, when treatment should best be given, and whether better treatments could be available. The SIOP-e NF-1 OPG working group aims for the next European protocol combined with registry (SAVING) to have standardized outcome measures, treatment indications and relevant randomisations to be able to answer these questions, with visual function as primary outcome measure.

With these aims, a multidisciplinary workshop to analyse radiological, clinical and visual factors in 83 patient cases (both treated and untreated) from 9 European SIOP-LGG 2004 centres was held in Nottingham in April 2014. There was no correlation between tumour outcome and visual outcome in the data. Consensus was achieved on visual outcome measures and classifying the tumour's anatomical involvement of visual pathways by modified Dodge classification (PLAN score). Agreed criteria for future case selection within trials were age, history of or documented visual decline, presence of severe visual symptoms, unreliable visual assessment, proptosis, and tumour involvement in the posterior optic radiations. The group has worked to refine these criteria into a visual risk assessment score and conducted a survey to investigate the degree of consensus on visual treatment indications in different patient settings.

[5] CONTACT LENSES VERSUS EYEGASSES AFTER PEDIATRIC CATARACT SURGERY

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This presentation focuses on the optical rehabilitation of aphakic pediatric patients. The optical management of this patient group is subject to a fast growth of the eye with subsequent radical refractive changes, while the visual system matures. The main concern is the risk of developing amblyopia if the visual input is unfocused or unequal between the two eyes. Based on a case the advantages and disadvantages of the use of respectively aphakic glasses and aphakic contact lenses will be discussed.

[6] SURGICAL STRATEGIES FOR THE CORRECTION OF APHAKIA IN CHILDHOOD

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Surgery for ectopia lentis and cataracts in childhood, especially in infancy, may result in secondary aphakia. Non-surgical management with glasses and especially with newer types of contact lenses can yield very rewarding results for the child and the family, but may not be a permanent solution due to non-compliance, intolerance or patient choice.

Surgical options for the correction of aphakia include secondary IOL implantation in the bag or sulcus, the anterior chamber or using iris, scleral (sutured or glued) fixation techniques.

All these techniques represent their own special intra- and post-operative challenges. The author's personal choice is sulcus or in-the-bag fixation in the presence of good capsular support, or retropupillary iris fixation using the Artisan lens in its absence.

[7] CHARACTERISTICS OF CHILDREN OPERATED FOR CATARACTS 2007-2016 RESULTING IN VISUAL ACUITY OF 0.8 AFTER 5 YEARS OF AGE - A REPORT FROM PECARE SWEDEN

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Aim: To investigate the factors associated with almost normal visual acuity results, defined as Snellen values of 0.8 or higher in the national Swedish Pediatric Cataract register, PECARE.

Methods: All eyes operated for cataract in Sweden before the age of 8 years, between January 2007 and July 2016, registered in the PECARE were included in the study. The monocular best corrected visual acuity of the eyes, (BCVA), was analyzed at follow-up visits at 5 and 10 years of age, respectively.

Results: A total of 504 eyes underwent surgery during the study period. Visual acuity was registered in 169 eyes at the age of 5 years and 13 eyes at the age of 10 years. A total of 10 %, (16/169) reached a BCVA of 0.8 or higher in the group of 5 years of age compared to 70 % (9/13) at the age of 10 years of age. Surgery was performed between 14 months and 5.9 years of age in the two groups with close to normal visual acuity. There was an improvement in visual acuity from age 5 to 10 in 38% of the eyes (5/13)

Conclusion: At the moment, the majority of the BCVA registered 10 years of age, in PECARE, have reached an almost normal visual acuity. For a substantial proportion of the BCVA registered at 5 years of age visual acuity improved from the age of 5 years to 10.

[8] IATROGENIC ADRENAL INSUFFICIENCY IS FREQUENT IN INFANTS TREATED WITH TOPICAL GLUCOCORTICOIDS AFTER OPERATION FOR CONGENITAL CATARACT.

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Local steroid is often used after eyesurgery, but little is known about the systemic effect of steroids applied to the eyes. We investigated 26 consecutive infants under the age of two years who were operated for congenital cataract. A postoperative standard ACTH provocation test was performed in all cases.

Of those tested during treatment with ocular steroids two thirds had suppressed adrenal gland function and two had clinical distinct features of Cushing syndrome. There was a significant difference in cumulated glucocorticoid dose per body weight given the last 30 days before the test in the suppressed patients as compared to the patients with normal test.

We recommend ACTH testing as part of clinical routine follow-up of children treated with ocular glucocorticoids.

[9] GLAUCOMA AFTER PAEDIATRIC CATARACT SURGERY

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Introduction

Glaucoma after paediatric cataract surgery is a well-known risk and a potential threat to vision. The aim of this paper is to present data on the occurrence of secondary glaucoma in children operated for cataract during eight years (2007-2014), in Sweden and Denmark, based on data from PECARE, a subdivision of the Swedish National Cataract Register,

Objectives

On 31 December 2014, a total of 678 operated eyes in 490 children were registered. Excluded were glaucoma pre-operative, uveitic or traumatic caused cataracts and eyes operated before 2007. The material thus consisted of 637 eyes in 239 boys and 215 girls (47%). 299 eyes had a registered follow up at one year of age, 322 at two years and 260 at five years.

Methods

Data was derived from the Paediatric Cataract Register (PECARE), a bi-national web-based surgical register. Children with a cataract extraction before 8 years of age between 2007 and 2014 were included.

Results

309 eyes (48.5%) had surgery before one year of age. Overall 105 eyes of 637 had a registered glaucoma (16.5%). Only 6 eyes of 328 who had surgery after one year of age had glaucoma. In 113 eyes with surgery before one month of age or small eyes (axial length <17mm and corneal diameter <10mm) glaucoma developed in almost 50% of the eyes.

Conclusion

Early surgery for cataract and small eyes increase the risk of early secondary glaucoma. Children who had surgery after one year of age rarely developed glaucoma.

[10] PARENTS OF CHILDREN WITH CATARACT - HOW DO WE SUPPORT THEM?

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Background and purpose

Having a child with cataract demands extensive self-management involving both the participation of and the active care provided by the parents. Being diagnosed with congenital or juvenile cataract starts with, but does not end with, surgery. It is a lifelong adjustment affecting not only the child but the whole family. Therefore, the purpose of this study was to gain knowledge on how to support these parents, in order to give their children, the best conditions to develop their vision and to have a good quality of life.

Methods

A qualitative descriptive design was used with open-ended in-depth interviews of 15 parents (5 mothers, 5 fathers, 5 couples) with a child diagnosed with cataract and operated on, to explore what their main concern is. What is it like to be a parent of a child with cataract? How can we support them? Grounded theory was used for analysis.

Results

Four concepts emerged; 1) Comprehension - deals with grasping and understanding what has happened. This seems to start right when the parents realize there is a visual problem. 2) Adherence to following the treatment - patching, eye drops, contact lenses which is central. 3) Facilitating – the parents are using many strategies to ease the way for their children and 4) Adjusting – several changes are made in everyday life.

Conclusions

Through this knowledge we can create guidelines for supporting parents with a focus on being resource oriented by enhancing their abilities, resources and capacities.

[11] AMBLYOPIA PREVALENCE AND RISK FACTORS IN CHILDREN WITH ANTERIOR POLAR CATARACTS

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Introduction

Congenital anterior polar cataract (CAPC) has been considered as a benign form for congenital cataract (CC), with a small risk of developing amblyopia. However, a clinical suspicion that the risk of amblyopia is high in these children led to the present study.

Purpose

To examine the risk of developing amblyopia in patients with CAPC and to identify risk factors for amblyopia.

Methods & Materials

The medical records for patients with CC that are currently being treated/examined at the Eye Clinic Rigshospitalet-Glostrup / Kennedy Center were reviewed. A total of 165 patients were found, of those 16 patients with CAPC were included. The study was approved by The Research Ethics Committees of the Capital Region of Denmark, Danish Data Protections Agency and Danish Health Authority.

Results

Twelve (75%) children had unilateral CAPC and 4(25%) had bilateral. Mean age was 3.5 years. The majority of children were girls (14 (88%)). Twelve (75%) were amblyopic and of those 7(58%) were anisometropic (>1 Diopter, (D), difference in spherical equivalent between eyes), 8(67%) had hypermetropia (>2 D), 1(8%) had myopia (>2.5 D), 5(41%) had astigmatism (>1 D) and 4(33%) had squint.

Conclusion

We found a high prevalence of amblyopia in children with CAPC. In the majority of those children, a refractive error seemed to be cause of amblyopia. The sample of this study is small and limited to children in active care for CC. We recommend that a larger population of children with CC are screened to provide a broader based estimate of amblyopia.

[12] TOPICAL BETA-BLOCKER TREATMENT FOR CAPILLARY HEMANGIOMA IN INFANCY

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The treatment of infantile hemangiomas changed from the use of corticosteroids to oral propranolol on the serendipitous discovery of propranolol's clinical effectiveness in 2008.

Since then, clinicians have begun to use topical beta blockers, in particular timolol maleate 0.5% gel forming solution, with good therapeutic effect, but fewer adverse reactions. Topical beta blockers are now used for lesions with both deep and superficial components and those that are amblyogenic. When initiated in the proliferative phase of the lesion, the effectiveness of the treatment can be seen within days.

Several illustrative cases of deep periocular and orbital involvement will be presented as well our immunohistochemical and electron microscopical findings.

[13] RETINOBLASTOMA

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Retinoblastoma (RB) occur in approximately 1 in 15 000 births. It is most often initiated by mutation of the *RB1* gene. The detection rate of a mutation is 95%. RB may be heritable or non-heritable, and involves one or both eyes. Untreated RB is fatal. In our countries, the survival rate is greater than 95%/5years. Early diagnosis and treatment are crucial. Leukocoria and strabismus are the most common presenting signs. During the last years, new treatments have been available and improved the outcome. Patients with heritable RB have a significant risk of secondary primary cancer.

[14] A CASE REPORT - INTRA-ARTERIAL MEPHALAN AS PRIMARY TREATMENT FOR RETINOBLASTOMA

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Retinoblastoma is difficult to treat due to the side-effects of high dose systemic chemotherapy when given to children. Local application allows administration of much higher doses, and accordingly intra-arterial chemotherapy delivered via the ophthalmic artery is emerging as a replacement of systemic treatment.

Two cases treated with this approach at Aarhus University Hospital are presented. The first patient, a 34 months old boy, was treated with the drug Mephalan three times, four weeks apart followed by adjuvant treatment of Transpupillary Thermal Therapy. The second patient, a 17 months old boy, has so far only been treated with Mephalan. In both cases the tumors has totally regressed and the eyes are so far preserved.

In conclusion, intra-arterial Mephalan appears to be safe and effective, and is likely to be used as primary treatment for retinoblastoma patients.

[15] RETINAL VASCULOPATHY

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[16] RETROBULBAR UNILATERAL ORBITAL ABSCESS SECONDARY TO DACRYOCYSTITIS IN A NEONATE WITH BILATERAL DACRYOCYSTOCELE

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Congenital dacryocystocele (CD) is normally a transient, self-resolving condition caused by nasolacrimal duct obstruction. CD occurs with a frequency of approximately 1/4000 births, most often in females and the majority of cases are unilateral. Acute dacryocystitis and periorbital cellulitis are rare complications to CD that require immediate intervention. Advanced infection such as retrobulbar orbital abscess secondary to dacryocystitis in a newborn is extremely rare.

We report a newborn boy with retrobulbar orbital abscess secondary to dacryocystitis facilitated by CD.

The infant presented with respiratory distress, sepsis and clinical signs of postseptal cellulitis.

The patient underwent bilaterally probing of the lacrimal ducts and intranasal incision of mucocoeles.

Intravenous Cefuroxim was administered before and after surgery. Three months after treatment the patient was well and without sequelae.

Dacryocystocele must be considered a differential diagnosis of newborns suffering from nasal respiratory difficulty.

[17] OCULAR FINDINGS IN INFANTILE MALIGNANT CLCN7-RELATED AUTOSOMAL RECESSIVE OSTEOPETROSIS

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Autosomal recessive osteopetrosis is a group of disorders characterized by generalized osteosclerosis and club-shaped long bones. Sclerosis of the skull base result in optic nerve compression, hearing loss and facial palsy. Absence of the bone marrow cavity results in severe anaemia, thrombocytopenia and hypocalcemia. Without treatment maximal life span is ten years but early (within two months of age) hematopoietic stem cell transplantaion (HSCT) can be curative.

A small subset of patients has a more severe disease with progressive neurodegeneration that progress despite successful HSCT. Retinal degeneration with macular and electrophysiological findings accompanies this condition. Two patients with infantile malignant *CLCN7*-related autosomal recessive osteopetrosis are presented with focus on their ocular manifestations, diagnostic work-up and implication for treatment.

[18] CORNEAL VASCULARIZATION, BLEPHAROPHIMOSIS, DEAFNESS AND ACROOSTEOLYSIS ASSOCIATED WITH MUTATIONS IN A NOVEL TYROSINE KINASE RECEPTOR

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Background

Our research group has examined hereditary forms of corneal vascularization to elucidate pathways involved in disease development. In 2006 Warburg and co-workers described a patient with a novel syndrome characterized by blepharophimosis, corneal vascularization, deafness, and acroosteolysis and noted his condition did not fit into any known syndromes. In 2013, Cinotti and co-workers reported a similar patient. The gene mutations associated with these overlapping phenotypes was not known.

Aim

The aim of this study was to investigate the molecular basis for this novel syndrome.

Methods and materials

Next generation sequencing was performed to identify the disease causing mutations. Patient and control fibroblasts were cultured from skin biopsies. Protein expression was examined using immunoblotting.

Results

Using next generation sequencing, we detected mutations in a tyrosine kinase receptor in both patients. One of the mutations results in an altered phosphorylation site of the receptor. Expression and activation levels of proteins downstream of the receptor vary between patient and control fibroblasts. Further clinical and molecular data will be presented.

[19] KARSCH-NEUGEBAUER SYNDROME

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Aim

Karsch Neugebauer is a rare syndrome, defined by congenital nystagmus, split hand and split foot anomalies. The syndrome affects about 1 in 90.000 babies. The syndrome is associated with additional abnormalities only rarely documented in the literature such as oral deformities. We want to report one of these rare cases with additional abnormalities.

Methods

Case report.

Results

One female case of Karsch Neugebauer syndrome, presenting with congenital horizontal nystagmus, ectrodactyly, optic nerve hypoplasia and corpus callosum abnormalities shown by MRI. Point mutation caused the phenotype where neither parent had the syndrome. A female offspring and her two sons also have ectrodactyly but no congenital nystagmus.

Conclusion

By reporting additional abnormalities in patients with Karsch Neugebauer syndrome we contribute to a better understanding of this condition. With only a few case reports in the literature, this case adds to the possible phenotypic presentations of this syndrome.

[20] HALLERMANN-STREIFF SYNDROME - A CASE REPORT

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Introduction: Hallermann-Streiff Syndrome (HSS) is a rare congenital condition characterized by dysmorphic facial features, eye- and teeth abnormalities.^{1,2} We report a case of the syndrome in a female, who was diagnosed at the age of 13 years.

Methods: retrospective review of the medical report regarding a Rumanian girl, who in 2015 moved to Denmark and shortly hereafter diagnosed with HSS.

Results:

Dysmorphic features. A beak-shaped nose, microstomia and mandibular retrognathia. Hypotrichosis and sparse supercilia. Microcephalic cranium with a wide forehead, parietal and frontal bossing. Height 132 cm (-5 SD), weight 48,3 kg (-0,7 SD).

Ophthalmological examination. BCVA: 0.1. Refraction: +12 at both eyes. IOP: 12/14 mmHg. Microphthalmic eyes, convergent strabismus and nystagmus. Microcorneas with band keratopathy. Old pigmented keratic precipitates. Deep anterior chambers without cells. Atrophy of the irises. Synechiae covering the pupils. Aphakia. Ophthalmoscopy revealed normal papillary configurations but cicatricial sequelae and degenerative areas of the retinas.

Dental examination. Severe oligodontia with 18 congenitally missing permanent teeth. The crowns of the maxillary central incisors were shovel-shaped. 14 deciduous teeth were still present.

Conclusion: This case is one of the few known cases of HSS in Denmark and demonstrates, despite all the main features of the syndrome, the possibility of spontaneous cataract resorption, which has been described in 8% of the previous published cases.³

References:

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² E. B. Streiff. Dymorphie mandibulo-faciale (tête d'oiseau) et alterations oculaires. Ophthalmologica, Basel, 1950, 120: 79–83.

³ London Ophthalmic Genetics Database (GENEEYE).

[21] REGIONAL DIFFERENCES OF THE INCIDENCE OF ROP IN SWEDEN OVER THE LAST DECADE INDICATE DISPARITY IN THE QUALITY OF NEONATAL CARE

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With the help of SWEDROP, national and regional incidences of ROP and frequencies of treatment in Sweden were evaluated from 2008 to 2015 as well as before and after implementation of new oxygen targets from 85-89% to 91-95%.

ROP was found in 31.9% of the 5734 infants with a GA < 31 weeks at birth and treatment was performed in 5.7%. There was an increased incidence of ROP during the eight-year study period, but no significant increase in frequency of treatment. Regarding the seven health regions, there were significant differences of both incidences of ROP and frequency of treatment.

[22] ETHICAL ISSUES IN NEONATAL CARE

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ROP is a problem in infants who is usually born extremely preterm and often have experienced several other significant perinatal complications. ROP is one of the four 'big' neonatal morbidities, the others being severe intraventricular haemorrhage/periventricular leucomalacia, necrotizing enterocolitis, and bronchopulmonary dysplasia. The number of these morbidities at the time of discharge from hospital predicts the risk of permanent moderate or severe neurodevelopmental impairment (NDI) in a linear fashion, reaching 60-70% when all four are present. A large fraction of the advantage of increasing gestational age at birth in terms of NDI appears to be due to the reduced risk of neonatal morbidity. Ethical dilemmas in this population specific to ROP is the late appearance, the stress and strains of screening and interventions for ROP - and perhaps the burdens of multiple neurosensory handicaps.

[23] PREDICTIVE FACTORS OF 6.5-YEAR OPHTHALMOLOGIC OUTCOME OF EXTREMELY PRETERM BIRTHS IN SWEDEN: A PROSPECTIVE POPULATION-BASED OBSERVATIONAL STUDY (EXPRESS)

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Background

A high prevalence of ophthalmologic abnormalities was found at 6.5 years' follow-up in a national cohort of children born before 27 gestational weeks. We aimed to analyze the prediction value of various pre- and postnatal risk factors as well as 2.5 year neurodevelopmental findings for the 6.5-year ophthalmologic outcome.

Design/Methods

A prospective population based study on long-term ophthalmologic outcome related to extreme prematurity. The participants comprised the Swedish cohort of children born at gestational age (GA) 22+0 to 26+6 weeks during 2004-2007. At the age of 6.5 years 399/486 (82%) survivors underwent ophthalmological examinations and were compared to a group of full-term children (n=300). Visual acuity, refraction and strabismus were evaluated and analyzed in time-lined regression models together with various pre- and postnatal risk factors as well as with the 2.5 year neurodevelopmental outcomes.

Results

The risk of abnormal ophthalmologic outcome increased with low GA (22-24 weeks odds ratio [OR] 1.9; 95% CI 1.1-3.1, when compared to 26 weeks GA), with severe retinopathy of prematurity (ROP) (OR 1.8; 95% CI 1.1-3.0) and with severe bronchopulmonary dysplasia (BPD)(OR 1.8; 95% CI 1.1-3.0). Brain injury was a significant risk factor at discharge from NICU, but not when adjusted to 2.5 year neurodevelopmental findings.

Conclusion

In extreme prematurity the most important risk factors for abnormal ophthalmologic outcome at 6.5 years of age were related to low GA, severe ROP, brain injury and BPD. These risk factors are important to consider in prediction of ophthalmological problems and in the clinical follow-up situation.

[24] VISUAL IMPAIRED CHILDREN IN DENMARK WITH FOCUS ON REHABILITATION

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In Denmark it is by law that children with visual impairment are registered in a national database. Children with visual acuity $\leq 6/18$, visual field < 20 degrees, hemianopia or proven retinal degeneration are all in the register. The children get access to spectacles, low vision aids and special education. New digital devices as smartphones and tablets are not yet a part of this and we have made a questionnaire regarding how they are used.

Results

In 2016 a total of 1798 children above the age of 2 years where know and of these 843 responded to the questionnaire. A total of 81% of the children above the age of 13 years responded, 48 % of those above the age of 6 years and only 27% of the youngest.

Tablets were used in all age groups: it was found that 88% of the all children had a tablet, 71% in the oldest age group had a smart phone, only 50% in the younger groups had smart phones, 20% had binoculars and 20% magnifying glasses, while 35% had special lightening. Many of the visually impaired children did not use their ordinary low vision aids but had replaced them with the new devices. They used many different apps.

Conclusion

There is no doubt that children can use the digital devices in order compensate for the reduced visual acuity and even children with multiple handicaps benefit from these. They replace the traditional low vision aids to some extent and should be regarded as eligible.

[25] VASCULAR ENDOTHELIAL GROWTH FACTOR (VEGF) INHIBITOR TREATMENT FOR RETINOPATHY OF PREMATURITY (ROP) IN HELSINKI UNIVERSITY HOSPITAL

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Purpose

Outcome of VEGF-inhibitor treatment for type 1 ROP.

Methods

Retrospective study of patients treated with VEGF-inhibitor for type 1 ROP disease between September 2011 and December 2016 in Helsinki University Hospital. The following data were recorded: gestational weeks (gw) at birth and at treatment, birth weight, classification of ROP, outcome (resolution of ROP, retinal vascularization, need for re-treatment, visual outcome). Indication for VEGF-inhibitor treatment was zone 1 or posterior zone 2 stage 2 plus, or worse, retinopathy of prematurity (ROP).

Results

17 eyes of 9 patients (mean \pm SD; gw 24.6 ± 0.9 and weight at birth 634 ± 67 g) were treated with intravitreal anti-VEGF agents (bevacizumab 11 eyes and ranibizumab 6 eyes). After treatment, incomplete peripheral vascularity was recorded in 9 eyes. Re-treatment with laser therapy was needed in 3 eyes with leakage on fluorescein angiography (FA), two of them having local retinal detachments. Incomplete vascularization was noted up to 31 months' post treatment. Visual acuities appeared normal for age in 11 eyes. Three patients had cortical visual impairment.

Conclusion

Long-standing follow-up and often investigation under general anesthesia including FA was needed. Vascular arrest in peripheral retina was frequently observed in our patients.

[26] EXTREMELY PRETERM CHILDREN HAVE REDUCED PHOTORECEPTOR FUNCTION

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Introduction

The aim of the study was to evaluate the retinal function with full-field electroretinogram (ffERG) in extremely preterm children aged 6.5 years and in a control group of children born at term.

Methods

The children were living in Uppsala County and participating in the national Extremely Preterm Infants in Sweden (EXPRESS) study. Binocular ffERG was assessed with the Espion Ganzfield System, using DTL electrodes in both eyes. Rod and cone stimulation with single flash strengths of 0.009, 0.17, 3.0 and 12.0 cd.s/m² together with cone stimulation 30 Hz flicker and single cone flash, 3.0 cd.s/m² were performed. The amplitudes and implicit times of the a- and b-wave were analysed.

Results

The preterm group had significantly lower amplitudes of the combined rod and cone responses (a-wave of 3.0 (p=0.003) and of 12.0 (p=0.017)) together with the cone responses of the 30 Hz flicker (p=0.026), when compared with the controls. The implicit time of the combined rod and cone responses of the a-wave of 12.0 was significantly longer (p=0.046) in the preterms, as well as of the cone responses (30 Hz flicker (p=0.043)).

Conclusion

Extremely preterm birth seems to affect the retinal function. FfERG revealed a dysfunction of the photoreceptors, which may contribute to the various visual problems in this new paediatric population.

[27] LONG-TERM OUTCOMES IN X-LINKED RETINITIS PIGMENTOSA IN DENMARK: COMPARISON OF PATIENTS WITH RPGR AND RP2 MUTATIONS

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Aim

To compare visual acuity and visual field outcomes between the two most common mutations in the two genes (RPGR and RP2) involved in X-Linked Retinitis Pigmentosa (XLRP), with debut in early childhood namely those in the Retinitis pigmentosa GTPase regulator gene (RPGR) and the Retinitis pigmentosa 2 protein regulator gene (RP2).

Methods

Retrospective analysis of longitudinal data collected between 1948-2014 in the Danish Retinitis Pigmentosa Registry. This retrospective analysis included the best-corrected visual acuity and Estermann visual field score from the right eye of all 84 male patients with XLRP in Denmark who belonged to one of 29 known pedigrees.

Results

A total of 1185 patient years were observed with a mean follow-up time of 14.6 years.

The RPGR mutation conferred a smaller annual BCVA reduction by 0.0156 ± 0.0075 logMAR compared with the RP2 mutation (p for interaction = 0.042). Conversely, the annual changes in Esterman scores did not differ between the mutations ($p = 0.80$).

Conclusion

This longitudinal study showed slower visual acuity loss in patients with RPGR compared with RP2 mutations and no difference in visual field loss, thus confirming the results of previous cross-sectional studies.

[28] INCIDENCE AND CLINICAL CHARACTERISTICS OF PEDIATRIC PERIOCCULAR DERMOID CYSTS

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Introduction

This population-based study was designed to report the incidence, clinical findings, and outcomes of periocular dermoid cysts diagnosed among children over a 20-year period.

Methods

The medical records of all patients 5 years of age or younger who were diagnosed with a periocular dermoid cyst from January 1, 1986, through December 31, 2005, were retrospectively reviewed.

Results

A total of 54 children were diagnosed during the 20-year period, yielding a birth prevalence of 1 in 638 live births. The mean age at diagnosis was 12 months and 29 (53.7%) were female. Forty-four (81.5%) occurred at the supratemporal orbital rim, 6 (11.1%) at the supranasal orbital rim, 3 (5.6%) in other periocular areas, and one (1.9%) within the orbit. Thirty-four (63%) had an ophthalmic exam, all without amblyopia or other ocular morbidity. Forty-eight (88.9%) patients underwent surgical excision with 7 (14.6%) having documented rupture of the cyst, none of whom had post-operative complications. Two (4.2%) patients were noted to have lesion recurrence; one at 8 months and the second at 1 year following surgery.

Discussion

Pediatric periocular dermoid cysts are uncommon lesions that generally occur in the first year of life in the supratemporal orbital rim. Most patients undergo uncomplicated surgical excision with rare recurrences.

Conclusion

The incidence of periocular dermoid cysts in this population is 1 in 638 live births. Complete surgical excision is the treatment of choice while ocular sequelae and postoperative recurrence are rare.

[29] INCIDENCE AND DE-NOVO MUTATION RATE OF MARFAN SYNDROME AND RISK OF ECTOPIA LENTIS

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Purpose

The purpose of this retrospective, observational clinical study was to investigate the incidence and de-novo mutation rate of Marfan syndrome and the risk for developing ectopia lentis.

Methods

The medical records of all patients newly diagnosed with Marfan syndrome (by the Ghent criteria) in Olmsted County, Minnesota, from January 1, 1976, through December 31, 2005, were retrospectively reviewed.

Results

Marfan syndrome was identified in 17 patients during the 30-year period, yielding an incidence of 0.52 per 100,000 people per year (95% CI, 0.27-0.77). The mean age at diagnosis was 24 years (range, 1 year to 51 years) and 9 (52.9%) were female. Five (29.4%) of the 17 were considered new mutations with a calculated mutation rate of $3.8 \pm 1.7 \times 10^{-5}$. Four (23.5%) were diagnosed with ectopia lentis, including 3 at the time of their Marfan diagnosis. Of the 14 patients at risk for developing ectopia lentis after being diagnosed with Marfan syndrome, 1 (7.1%) developed it during a mean follow-up of 9 years (range, 0 to 26 years). Twelve (70.5%) of the 17 were diagnosed with a dilated ascending aorta during a mean follow-up of 11 years (range, 6 months to 29 years).

Conclusions

The incidence and de-novo mutation rate of Marfan syndrome in this population-based cohort was higher than prior reports. The prevalence of ectopia lentis, not previously reported, occurred in approximately one-fourth of the study patients, and more commonly around the time of the initial Marfan diagnosis.

[30] VISUAL ACUITY AND VISUAL PERCEPTION IN ADOLESCENTS WITH SURGICALLY TREATED HYDROCEPHALUS - A LONG-TERM FOLLOW-UP

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Background

The incidence of congenital hydrocephalus (HC) is estimated to approximately 0.7-1.1 cases per 1000 live births. Symptoms vary; cerebral palsy and epilepsy are common as well as visual deficits. Almost 60% have visual perceptual problems (VPP) which means difficulties with interpretation of visual input.

Aims

To study visual acuity (VA) and VPP over time in a group of individuals with HC surgically treated in infancy, and compare the results with a healthy control group. Furthermore, to investigate whether there is a correlation between VA and VPP or not.

Methods

Twenty-six adolescents (10 females, 16 male), born 1999-2002 (median age 15.0 years), with HC were studied. Visual acuity was tested and VPP were evaluated through structured history-taking. The results were compared with data from participants' childhood (median age 8.7 years) and with an age- and sex matched control group (n=31).

Results

Fifteen out of 23 (61%) adolescents reported VPP, 12/28 (43%) in childhood (n.s.) and 2/31 (6.5%) in the control group ($p < 0.0001$). The median VA in childhood was 0.9 decimal (fix-1.25) and 1.0 decimal (fix-1.25) in adolescence compared with 1.25 (1.0-1.25) in healthy controls ($p < 0.0001$). No correlation between VA and VPP was found.

Conclusion

Our results indicate that children with HC do not develop significantly more or less VPP in adolescence. Approximately 60% reported VPP, compared with 6.5% in healthy controls. A wide range in VA was noted – some adolescents with HC could only fixate while others had a normal VA. However, no correlation between VA and VPP was found.

[31] IPAD OR PATCHING FOR AMBLYOPIA

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A binocular approach to treating anisometropic and strabismic amblyopia has recently been suggested using iPad technology instead of conventional patching. This presentation will summarize the background of the potential role for binocular treatment and explore the results of two just-completed randomized trials comparing part-time patching with iPad binocular treatment. The importance of compliance will be discussed. The design and rationale for a just-launched new randomized trial of alternative iPad binocular treatment will be presented. A current evidence-based algorithm for amblyopia treatment will be outlined.

[32] PREVALENCE OF AMBLYOPIA AND VISION SCREENING RESULTS - A NORDIC OVERVIEW

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Purpose

To establish the distribution of the prevalence rates of residual amblyopia in vision-screened Nordic populations and the effectiveness of vision screening and amblyopia treatment.

Material

Data is based on cross sectional epidemiological prevalence studies conducted by the authors and a thorough literature review on previously published epidemiological Nordic research on amblyopia.

Methods

The participants of the included studies underwent clinical ophthalmological examination followed by variable degree of supplemental orthoptic examination.

Results

Published epidemiological data on amblyopia before and after vision screening was available from Denmark, Finland, Norway and Sweden. The definition of amblyopia varied between studies. Prevalence data on amblyopia in previous non-screened not treated Nordic populations varied between 2.9% -3.2%. The prevalence data on residual amblyopia, defined as best corrected visual acuity (BCVA) in worse eye of ≤ 0.6 decimal, was found in 1.1% of the Swedish population. This was similar to 1.2% found in the Finnish population using the same definition. Using a more restrictive definition of BCVA in worse eye ≤ 0.5 lowered the prevalence to less than 1% in both Danish and Swedish populations.

Conclusion

Vision screening has significantly lowered the prevalence of amblyopia in Nordic countries. Results for residual amblyopia and effectiveness of visual screening and amblyopia treatment are very similar in comparable studies of Denmark, Finland and Sweden. We suggest future Nordic studies to use same international definition of amblyopia to enable precise comparison.

[33] VISION SCREENING IN CHILDREN 5 TO 8 YEARS OLD LIVING IN THE REGION OF VÄSTRA GÖTALAND, SWEDEN - A PROSPECTIVE, MULTICENTRE STUDY

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Introduction

Vision screening in 4 year olds has been performed in Sweden since the 1970s and is also recommended in schoolchildren aged 6-7 years. In Region Västra Götaland (VGR), a visual acuity (VA) of 0.8 decimal is the referral cut-off in children aged 5-7 years. The aim was to evaluate the current referral criteria in VGR with 1.6 million inhabitants.

Methods

During the period Oct 2014 to June 2015, all children at age ≥ 5 years < 8 referred from primary health care centres and schools to the four main referral centres in VGR, with VA of 0.65 decimal but not better in one or both eyes, were included. At the eye clinic VA with KM chart 3 m, refraction in cycloplegia, eye motility, cover test, stereo test and a slit lamp examination were performed.

Results

259 children (139 female) were included. Median age at referral was 5.7 years (5.0–7.8 years) and at examination was 6.1 years (5.2–8.2 years). 32% of the children were prescribed glasses due to subnormal VA with refractive errors and/or asthenopic problems. **Heterophoria was found in 22%; no one had heterotropia.** Amblyopia was found in 6 (2%) children. After prescribing glasses, and in one case also occlusion, the VA became normal in all amblyopic children.

Conclusion

A screening program with a cut off screening limit of 0.8 decimal is accurate and covers the children needing glasses due to refractive errors. All amblyopic children received normal VA at follow-up. Education of those performing the screening is important.

[34] CURRENT AMBLYOPIA SCREENING AND TREATMENT STRATEGIES IN FINLAND 2017

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In Finland screening for amblyopia is carried out as a part of routine visits at the child health clinics. Public health nurses evaluate visual acuity using a standardized test at the age of 3 and 4 years. Visual acuity is also checked at 5 and 6 years of age, if needed. In addition, at general practitioner's visits (at the age of 4-6 weeks; 4, 8 and 18 months, and 4 years) eye contact and fixation, external eye structures, red reflex, grasping, and eye alignment (a Hirschberg lamp test and cover test) are evaluated. There are no official national guidelines on how amblyopia should be treated and there are no studies on how Finnish ophthalmologist treat amblyopia. The results of a recent questionnaire on amblyopia treatment strategies are discussed.

[35] VISION SCREENING IN SCHOOL STARTERS

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Abstract not available at the time of print.

[36] HONORARY LECTURE: VISUAL IMPAIRMENT IN A 100 YEAR PERSPECTIVE

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A hundred years ago the situation for the visually impaired children was very different from the current circumstances. The most common disorders causing the visual impairment were corneal scarring, sympathetic ophthalmia, optic nerve atrophy and pediatric cataract. Education and work opportunities were different and more limited. The changed situation during the past century will be discussed as well as recent research resulting in improved care of patients. Visual impairment in children can in most cases not be treated or avoided with the exception of ROP and pediatric cataract. Therefore, optimised care of these patients is essential and quality registers can be a resource in our endeavours.

[37] NEUROANATOMY: 4TH NERVE, SUPERIOR OBLIQUE MUSCLE AND OCULAR MOTILITY

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The trochlear nerve (or the fourth cranial nerve) is a pure motor nerve; its only function is to innervate the superior oblique muscle of the eye. It is the smallest and at the same time the longest of the cranial nerves. The trochlear nerve is the only cranial nerve that completely cross over to the opposite side, and also the only nerve that leave the brainstem from the dorsal side. The anatomy of the superior oblique muscle is special, as the direction change radically as the tendon runs through the bony trochlea. The different functions of the muscle will be discussed.

[38] DIAGNOSIS OF 4TH NERVE PALSY

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Diagnosis of 4th nerve palsy will be discussed, including symptoms and signs of congenital and acquired palsy, as well as of congenital and acquired palsy. Examination technique and testing of cyclodeviation will be reviewed.

[39] INTRACRANIAL PATHOLOGY AND 4THNERVE PALSY

Darius Hildebrand¹

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Congenital anomalies of the trochlear nerve are the leading intracranial pathology of 4th nerve palsies. Thin-section, high-resolution 3-Tesla MRI has demonstrated absence of the ipsilateral trochlear nerve and variable degrees of SO hypoplasia in 73% of 97 patients with congenital 4th nerve palsy.

The second most common intracranial pathology, and leading cause of all acquired 4th palsies, is intracranial trauma, often accounting for bilateral palsies due to the proximity of the trochlear nuclei, fascicles and nerves in the dorsal mesencephalic brainstem. The 4th nerve's proclivity for trauma is due to its special anatomic features (it is the smallest and longest of the ocular motor nerves and its propinquity to the tentorial edge) and hence vulnerability to coup-contrecoup contusional and avulsive injuries along its anatomic course.

Among children, congenital and traumatic causes account for over 92.7% of cases in an American population-based study and 70-95% in tertiary hospital-based series.

Among adults, microvascular causes are the third most important cause.

Hydrocephalus, neoplasm, meningitis, aneurysms, infarction and haemorrhage are rarer, but important treatable other causes.

Despite all investigations, no clear intracranial pathology will be found in a residual group.

[40] SURGICAL MANAGEMENT OF 4TH NERVE PALSY – AN OVERVIEW

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Fourth nerve palsy leads to a row of signs and symptoms such as vertical diplopia with torsion, head tilt and neck pain. Objective findings of under -and overacting extraocular muscles with vertical squint and cyclotorsion of the eyes are found.

An overview to understand the surgical procedures used will be given in order to achieve the goal of a patient –free of diplopia in almost all positions of gaze, with binocular single vision and stereopsis. Normal head position without neck pains, reversal of the cyclotorsion and parallel eye axis, can be achieved after a single, combined or additional surgical procedures.

[41] INFERIOR OBLIQUE WEAKENING PROCEDURES

Juho Pekkarinen¹

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Surgical management of inferior oblique overaction is based on weakening or changing the function of the inferior oblique muscle. The most commonly used techniques include myectomy, recession and recession with anteriorization.

[42] SUPERIOR OBLIQUE TUCK PROCEDURES

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Abstract not available at the time of print.

[43] VERTICAL RECTUS MUSCLE RECESSIONS IN 4TH NERVE PALSY AND THE USE OF TOPICAL ANESTHESIA +/- ADJUSTABLE SUTURE

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Purpose is to highlight benefits and indications for topical anesthesia in strabismus surgery. Special considerations using pure topical anesthesia in vertical rectus recession surgery will be given. Principles for special surgical technique during vertical rectus muscle recession with topical anesthesia, to avoid pain and obtain good results, will be explained in details. Finally, preliminary outcomes of a cases series of vertical rectus muscle recession using topical anesthesia in 4th nerve palsy will be presented. Results are evaluated by patient tolerance, i.e. peroperative pain score scale, and surgical outcome.

[44] PRELIMINARY RESULTS OF EARLY SURGERY (AGE < 2 YEAR) FOR CONGENITAL ESOTROPIA

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Abstract not available at the time of print.

[45] TUCKING OF PARETIC OCULAR MUSCLES AS A RELATIVELY UNTRAUMATIC METHOD TO ACHIEVE BETTER OCULAR ALIGNMENT AND MOVEMENT

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The usually preferred method of achieving better alignment and movement when dealing with a paresis of extraocular muscles is a transposition of muscles that retain their function, i.e. lateral transposition of the Superior and Inferior Recti.

I have found these transpositions disappointing both with regard to impact on alignment and movement as well as side effects such as induced height deviations and late scarring.

As an alternative I have used a large tuck of the paretic muscle, which is a simple approach with less side effects and a minimal impact on blood supply. It can also easily be augmented in a subsequent operation.

[46] AN UNUSUAL INCIDENT AFTER STRABISMUS OPERATION

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Our patient is a normally fit and well seven-year-old girl with esotropia and high AC/A hyperopia. An overacting left inferior oblique muscle was also noted. After an uneventful bilateral strabismus surgery; recessions of both medial recti with posterior fixation sutures and a recession of the left inferior oblique, the recovery became complicated. Orbital MRI was carried out and it revealed an undesirable finding. The course of events will be described in detail.

[47] SCLERAL IMPLANTS IN STRABISMUS SURGERY ON GRAVES' ORBITOPATHY PATIENTS

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Abstract not available at the time of print.

[48] DIPLOPIA CAUSED BY ACQUIRED SYNKINESIS OF OCULOMOTOR AND MASTICATION MUSCLES?

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A young woman who had a congenital ptose in her right eye but otherwise normal visual status as a child developed troublesome diplopia when eating at the age of 16.

The case is presented, including a video. Causes of acquired synkinesis are briefly discussed.

The audience is invited to suggest further examinations of the patient or possible treatment of the diplopia.

[49] FOUR STRAIGHT MUSCLE SURGERY ON THE SAME EYE

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Abstract not available at the time of print.

[50] TRAUMATIC ABDUCENS NERVE PALSY

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48-y/o male with a serious accident at work (fallen into a 3.5 high sewer trench → ICU) one year earlier. Left orbital floor fracture → Titanium plate. Minor right orbital wall fracture → no repair needed. No muscle entrapments. Severe right abducens nerve palsy and a slight hypertropia ← no recovery over a year. VA 25/20 (OD) and 30/20 (OS). Hertell EOM 13/99/12mm. Normal SFL. Ta 10/15. BM: No special findings. Prism-Cover-Test (PCT): +110 Prd+ VD 5-12 PrD far and +100 PrD near. Head turned and slightly tilted.

I Operation: Transposition totalis (Superior and Inferior Rectus OD) with augmentation+ Botulinum toxin to Medial Rectus (OD)

2M post op: Patient very happy:

PCT: +12 Prd far and +12Prd-VD2Prd near. Head almost straight with a slight chin down intermittently, no double vision.

BUT 10M post op: ET/abducens nerve palsy increased again:

	+35PrD	
+75 PrD-VD10 PrD	+45PrD – VD 6 PrD	+40 PrD
	+75PrD-VD 6 PrD	

And in near: +50 PrD –VD6 PrD; 19 degrees excyclotorsion, V-syndrome (sdr), small left hypertropia + abducens paresis (OD)

II operation: Harada Ito (OS)+ Medial Rectus recession and hypooponation OU (4.0/3.0 OS and 6.0/3.0OD). The patient will be examined on the 3/2017 and the final results will be reported in the oral presentation.

Points:

- a case of traumatic abducens nerve palsy
- the use of Botulinum toxin (although with a temporary effect)
- advanced operations: Transposition totalis, Harada-Ito and correction of V-sdr