OM OPTIKUSHYPOPLASI
MED FOKUS PÅ UTVECKLINGSAVVIKELSER

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Synskadeorsaker förr och nu

- För 100 år sedan; infektioner, undernäring och olyckor (Norrie, 1927)
- För 50 år sedan; congenital cataract, retinoblastom och ROP (Lindstedt, 1972)
- För 20 år sedan;neuro-ophthalmologiska sjukdomar (50%), CVI, ONH, synnervsatrofi.
- Prenatala skador 64%.
- CVI i 25 % av fallen. Ca 3000 barn ingår i rapporten (Blohmé och Tornqvist, flera publikationer)

Få barn idag är ”bara” synskadade

- I det svenska synskaderegistret (1994) hade 60 % additional impairments (mental, motor or hearing)
- Hälften hade neuro ophtalmologiska sjukdomar-88 % tilläggshandikapp
- I CVI gruppen hade 96 tilläggshandikapp
- Färre barn föds idag blinda men fler har synrester + andra funktionshinder

Varför denna ökning?

- Fler svårt sjuka barn överlever
- Fler mycket prematurfödda barn överlever
- Synskadan uppmärksammas oftare hos barn med andra ”major handicaps” (Warburg 1970; hälften av alla barn ID har också en synskada)
- Nya och mer effektiva diagnosmetoder
- Den har fått ”ett namn”

Ett förändrat panorama i forskningen om konsekvenserna av synskadan

- Sedan -50 talet har fokus legat på förlusten/avsaknad av syn i relation till (social)utveckling (Freiberg,Warren, Preisler m.fl)
- Från 90-talet ligger fokus på ”brain and brain lesions” (Cass, Sonksen, Dutton, jan, Freeman, Jacobson, Ek och många fler) spec. kognitiv utveckling

Blindhet och autism

Är idag en vanlig kombination

Ca. 30% i en heterogen population (olika synskadeorsaker) av barn och ungdomar med blindhet.

Mycket vanligare vid vissa specifika diagnoser speciellt ROP och ONH

Lite´historik

- 1943. Kanner L. Autistic disturbances and affective contact
- 1944: Asperger H. Die ”autistishen psychopathen im Kindesalter”
- På 40-talet kom kuvöserna..

- Keeler 1958, ROP och autism.

I en studie av barndomsschizofreni, 5 blinda barn med ROP;

”presented the most strikingly similar picture to infantil autism”

- Blank 1959, ROP och autism.

Liknande fynd

- Bender & Anderman 1965,ROP och autism

22 barn blinda pga ROP. ”brainlesions related to intrauterine factors”

- Glavin 1966, ROP och autism

”drastic changes in oxygen pressure neonataly”

Joan Chase (1972)

studied the relationship between several background variables and the diagnosis of autism in 263 children with ROP, as well as the effects of early maternal deprivation. The only relations found to be significant were “those concerning the neurological sequelae due to low birth weight, short duration of gestation and high levels of oxygen over a long period”.

This ended the discussion for a long time!

Synen på autism

- In 1960, Selma Fraiberg established a guidance service for blind children in New Orleans.
- 7 of 27 children in this group ”presented a clinical picture that very much resembled autism in sighted children”
- brain damage was discussed as one possible cause, but she used the term “ego-deviations” to describe the condition (Fraiberg & Freedman, 1964).
- Jämför Bruno Bettelheim ”refrigerator mothers”
- Alltså kontaktstörning, inte en kognitiv störning!

Nyare studier
ROP och autism, 1998

- Relation between blindness due to retinopathy of prematurity and autistic spectrum disorders.
  Ek, Fernell, Jacobson & Gillberg 1998. *Dev. Med.*

- 27 barn f. 1980-90. Blinda pga ROP
- 15 hade autism
- 4 hade autistic like condition
- 19/27=2/3
- Mental retardation, epilepsi, CP och hydrocephalus förekom mycket ofta.

Table I: Clinical follow-up data on 27 children with ROP stage 5

Nr	*Sex*	*Intelligence level*	*Epilepsy*	*CP*	*IH*	*AD/ALC*	*CARS*
1	F	SMR	+	–	+	AD	50
2	F	SMR	–	–	–	AD	49
3	F	SMR	–	+	–	AD	35
4	F	SMR	–	+	–	AD	55
5	M	SMR	+	+	+	AD	39
6	M	SMR	–	+	–	AD	50
7	F	SMR	–	+	+	AD	36
8	F	SMR	–	–	–	AD	47
9	F	MMR	+	+	+	ALC	32
10	M	MMR	+	–	–	AD	50
11	M	MMR	+	+	–	ALC	[b]
12	M	MMR	–	–	–	ALC	25
13	M	MMR	–	–	–	AD	43
14	M	MMR	+	+	–	AD	39
15	F	MMR	–	–	–	AD	35
16	M	MMR	–	–	–	AD	47
17	M	MMR	–	–	–	AD	33
18	M	MMR	–	–	–	AD	36
19	F	Normal	–	–	–	[a]	–
20	F	Normal	+	–	–	[a]	–
21	F	Normal	–	–	–	[a]	–
22	M	Normal	–	–	–	[a]	–
23	M	Normal	–	–	–	[a]	–
24	F	Normal	–	–	–	[a]	–
25	M	Normal	–	–	–	ALC	30
26	F	Normal	–	–	–	[a]	–
27	M	Normal	–	–	–	[a]	–

CP, cerebral palsy; IH, infantile hydrocephalus; AD, autistic disorder; ALC, autustic-like disorder; MMR, mild mental retardation; SMR, severe mental retardation; [a]fulfilled 2–3 CARS, Childhood Autism Rating Scale criteria of the DSM-IV; [b]information incomplete.

Beteende

- Tillbakadragna och reserverade
- Försiktiga och långsamma
- Inte så mycket utbrott
- Nästan alla talade i rel. normal tid och utvecklade ett bra språk!
- Kommunikationen brast dock
- Extremt upptagna av ljud

Föräldrarnas berättelser

- Fas 1; överlevnad, sjukdom, ögonoperation och dramatik
- Fas 2; anpassning till att ha ett blint barn
- Fas 3; utveckling, är han normal?
- Fas 4; mycket personal ”developmental lag theory”
- Fas 5; nya diagnoser, tiden har gått

Lebers cong. amauros

- Steinberg et al 1992.Få avvikelser från andra blinda barn.
- Casteels et al 1996. 11/14 normal utveckling, normal CT.
- Rogers 1989 och Goodman 1995. Enstaka fall av autism.
- Black& Sonksen 1992.Enstaka fall av CNS inslag=beteendeproblem, utvecklingsstörning.
- Blohmé&Tornqvist 1997.Inga rapporterade tilläggshandikapp hos LCA.

Fazzi et al. 2007. LCA: is there an autistic componet? *Dev. Med.*

- 24 barn undersöktes.
- 23-inga egentliga neurologiska avvikelser
- 4/24 visade en måttlig-mild grad av autism
- Slutsats-” no common neurological damage underlying both the visual deficit and the autistic disorder was confirmed”

Autism in visually impaired individuals. Mukkades et al 2007. *Psych and Clin neurorosc .*

- 257 blinda barn 7-18 år. Olika ophthal. diagnoser.
- 30 av dessa- autism
- ”regardless the type of ophthalm. problem, brain damage/dysfunction has an important effect contributing to autism”
- ”severity of VI, brain damage and mental retardation are the likely factors to account for autism..

Cognitive and behavioural characteristics in blind children with bilateral optic nerve hypoplasia**.**

Ulla Ek,, Elisabeth Fernell, Lena Jacobson. Acta Paed. 2005

- Data från barnläkarjournaler, föräldraintervjuer, lärare, upprepade psykologutredningar av 13 blinda barn med ONH.
- Födda 1988-98.
- Alla hade medellinjesmissbildningar visat genom neuroimaging och/eller hormonella avvikelser.

ONH-opticushypoplasi

Liten och underutvecklad synnerv: En av de vanligaste orsakerna till blindhet hos barn i västvärlden.Förekomsten har ökat under de senaste två decennierna, delvis beroende på att man nu är bättre på att ställa diagnos. Dessa barn har förutom sitt synproblem ofta hormonella och neurologiska problem.

Table1. Paediatric and imaging data.

Child	GA	Neonatal period	Cerebral imaging	Hormonal deficiencies
1	39	hypoglycemia jaundice	CT/absent SP	GH, ACTH, TSH
2	42	hypotonia hypoglycemia	MRI/absent SP	GH
3	40	uneventful	MRI/absent SP	0
4	42	hypotonia hypoglycemia jaundice	MRI/normal	GH, ACTH, prolactin, ADH
5	38	hypoglycemia jaundice	MRI/absent SP and pituitary stalk, ectopic poster. pituitary	GH, ACTH
6	42		MRI/ectopic.poster.pituitary	GH
7	42	hypoglycemia jaundice	MRI/absent SP, arachnoid cyst-cistern ambiens right	GH, ADH, ACTH
8	41	hypotonia hypoglycemia	MRI/ absent SP and pituitary stalk, ectopic poster. pituitary	GH, ACTH, TSH
9	40	uneventful	MRI/absent SP and pituitary stalk, ectopic poster. pituitary	GH, ADH
10	42	uneventful	MRI/holoprosencephaly, dysgenesis corpus callosum	GH
11	40	hypoglycemia hypotermia jaundice	MRI/absent SP	GH, TSH, ACTH
12	-	born abroad	CT/normal	GH, ADH
13	39	uneventful	CT/absent SP, schizencephaly right	0

GA: gestational age; CT: computerized tomography; MRI: magnetic resonance imaging; SP: septum pellucidum; GH: growth hormone; TSH: thyroid stimulating hormone; ACTH: adrenocorticotropic hormone; ADH: antidiuretic hormone.

Table 2. Cognitive and behavioural characteristics.

Child	Early behaviour characterics	Cognitive level	Number of cognitive assess-ments and methods.	Autism Spectrum Disorder	Age of autism assessments
1	tantrums	MMR	2 [1, 2, 3]	autism	4y; 8y
2	tantrums	MMR	3 [1]	autism	5y; 6y: 7y
3	tantrums	normal	3 [3]	autism	10y;12y
4	tantrums	near normal	2 [1, 2]	autism	5y; 6y
5	tantrums	normal	3 [1, 3]	autism	5y; 10y
6	screaming	MMR	2 observation	autism	5y ,6y
7	tantrums	MMR	2 [2]	ALC	6 y; 8 y
8	extremely stubborn	normal	1 [2]	ALC, ODD	6y
9	tantrums	normal	2 [1, 2, 6]	ALC	6y
10	-	normal	1 [3, 4, 6]	no	
11	tantrums	normal	2 [1, 3, 4]	no	
12	passive	MMR	2 [1, 3 ,5, 6]	no	
13	tantrums	normal	2 [3, 6]	no	

ODD – oppostional defiant disorder ; MMR- mild mental retardation.
Methods for cognitive assessments:Griffiths [1] ; WPPSI [2] ; WISC III [3] ; ITVIC [4] ; Reynell Zinkin Scales [5] ; NEPSY [6] ;
observation - could not participate in a testing procedure, a structured observation was performed instead.

Behavioural characteristics

- Severe mood swings and temper tantrums were common, especially during the first years of life. Later in life sluggish tempo, low frustration tolerance and a narrow range of interests were common
- All children could talk and 12 /13 had started to talk at the expected age, but their language had clear deficiencies with respect to communicative ability. Their superficially fluent language seemed to mask the underlying problems with coherence, contexts and the meaning of situations encountered in everyday life situations.

- OBS- talet!!!

Neurodevelopmental Disorders in Optic Nerve Hypoplasia: A Population-based Cohort Study

Sara Dahl, MD, Ronny Wickström, MD, PhD, Ulla Ek, PhD, Kristina Teär Fahnehjelm, MD, PhD

Study group

- 65 children and youth < 20 years in Stockholm county dec 2009
- 35 bilateral, 30 unilaterala, 51% girls
- 53 neurologically ochophthalmologically assessed

Cognitive results

Resultat

- ID (mental retardation) in 20/55 (36 %)

Bilat ONH: 18/32 (56%)

Unilat ONH: 2/23 (9%) P< 0.001

Autism

Results

- Autism in 7/42 (17%)

  Bilat ONH: 5/21 (24%)

  Unilat ONH: 2/21 (10%) P> 0.05
- ADHD in 5 patients (3 unilat, 2 bilat)

Children with blindness – major causes, developmental outcomes and implications for habilitation and educational support: a two-decade, Swedish populationbased study.de Verdier K, Ek U, Löfgren S, Fernell E

- Included;Birth years between 1988 – 2008. Pre/perinatal or early infancy blindness category 4 – 5 (WHO), i.e. total blindness or light perception
- Search procedure: Medical, psychological and pedagogical files and records were reterieved from The National Agency's two national resource centre units, and central archives

- Phase 1: Files of every single child with VI born 1988 – 2008 were examnined

- Phase 2: An in-depth search of the files of all children who matched the inclusion criteria was performed
- Totalt 150 barn, 80 flickor, 70 pojkar

	Girls (n=80)	Boys (n=70)	Total (n=150)
Prenatal antechiasmal causes (n=93)			
Optic nerve hypoplasia (ONH)	9	14	23
Leber congenital amaurosis (LCA)	13	9	22
Optic nerve atrophy (ONA)	8	8	16
Microphtalmia	5	4	9
Anophtalmia	6	1	7
Congenital glaucoma	2	2	4
Retinal dystrophy	2	0	2
Coloboma	1	0	1
Incontinentia pigmenti	1	0	1
Norrie disease	0	1	1
Persistent hyperplastic primary vitreous (PHPV)	0	1	1
Sclerocornea	1	0	1
Unspecified eye malformations	3	2	5
Prenatal retrochiasmal causes (n=10)			
Various cerebral malformations	6	4	10
Peri-/postnatal antechiasmal causes (n=42)			
Retinopathy of prematurity (ROP)	14	22	36
Retinoblastoma	3	0	3
Congenital cataract	1	1	2
Optic glioma	0	1	1
Peri-/postnatal retrochiasmal causes (n=2)			
Cerebral damage due to early trauma or disease	2	0	2
Cause not defined whether pre/peri- or postnatal (n=3)			
Unspecified eye disease (antechiasmal)	2	0	2
Unspecified tumour (retrochiasmal)	1	0	1

Cerebral involvement and additional disabilities

- Only blindness 22% (n=33)
- Diagnosed cerebral involvement 33% (n=50)
- Epilepsy 18% (n=27)
- Additional impairments 72% (n=108)
  - Intellectual disability (ID)
  - Autism Spectrum disorder (ASD)
  - Motor impairment
  - Attention deficit/hyperactivity disorder (ADHD)
  - Hearing impairment
- 54% (n=81) had more than one additional impairment
  - ASD + ID
  - ID + motor impairment
- Unknown 6% (n=9)

Prevalence of autism in present study

- 31% (n=47) met the criteria for autism/Asperger/PDD-NOS
- 38% (n=57) if prominent autistic features were also included in the ASD-spectrum
- 63% (36/57) also had ID
- Dominating causes of blindness in the ASD-group:
  - ROP 37 % (21/57)
  - ONH/SOD 28 % (16/57)
  - LCA 14 % (8/57)

Prevalence of ASD in specific aetiological subgroups

- In the ONH-subgroup 70% had ASD
- OBS 100% av SOD- full autismdiagnos

- In the ROP-subgroup 58% had ASD

- In the LCA-subgroup 36% had ASD

Alltså..

- Blindhet ***per se*** leder inte utvecklingsavvikelser och/eller autism.
- Blindhet i kombination med skador och missbildningar av CNS; stora risker för utvecklingsavvikelser och/eller autism.
- Ordentlig utredning av barnet **och** av miljön är absolut nödvändigt!!