

Ågrenskas dokumentationer familjevistelser

Diagnos	Årtal
1p36-deletionssyndromet	2013
4p-deletionssyndromet/Wolf Hirschhorns syndrom	2020, 2012
16p11.2 deletion och duplikation	2019
22q11 deletionssyndromet	2000, 2002, 2005, 2007, 2015, 2018, 2022, 2023 (lättläst version)
Adhd och damp hos flickor	2000
Adrenogenitalt syndrom	2000, 2001
Aicardis syndrom	1998, 2002, 2010, 2017
Akondroplasi	1998, 2003, 2005, 2008, 2018
Albinism	2001, 2008
Alströms syndrom	2004
Analatesi	2006, 2022
Angelmans syndrom	1999, 2001, 2008, 2012, 2015, 2019, 2023
Anonymous	2000, 2003, 2006, 2012, 2015
Anorexia nervosa	1994
Aperts syndrom	2001, 2008, 2013, 2018
Artrogypos (AMC)	1996, 1999, 2001, 2005, 2007, 2008, 2013, 2016, 2023
Aspartylglykosaminuri (AGU)	2010
Aspergers syndrom	1997 (syskon), 1999, 2002, 2003
Ataxia telangiektasia	2005
Autism	1996
Autoimmuna encefaliter	2018
Bardet-Biedls syndrom	2019
Barnreumatism	2000
Beckers muskeldystrofi	2002, 2009, 2016, 2024
Beckwith-Wiedemanns syndrom	2000, 2008, 2015, 2023
Blåsexstrofi	1998, 2002, 2022, 2023
Blödarsjuka	1996, 2007
Bronkopulmonell dysplasi	2007
CDG-syndromet	1996, 2004, 2014, 2022
Charcot-Marie-Tooths sjukdom	2013
CHARGE-syndromet	1996, 2006, 2017, 2024
Cilieimmobilitetssjukdomar	2000
Coffin-Siris syndrom	2021
Cornelia de Langes syndrom	2020

Diagnos	Årtal
Corpus callosum agenesi	1995
Cri du chat syndrom	1996 (anhöriga) 2000, 2005, 2011
Crouzons syndrom	2001, 2023
Cystinos	2002, 2017
Cystisk fibros	1999, 2003, 2007, 2009, 2013, 2014
Cytomegalovirus, CMV	2001, 2007, 2013
Diamond-Blackfans anemi (DBA)	1995, 2006
Dravets syndrom	2011, 2014, 2017, 2020
Duchennes muskeldystrofi	2014, 2015, 2017, 2018, 2019, 2020, 2022
Dyskinetisk cerebral pares	2002, 2008, 2012, 2023
Dysmeli	2000, 2002, 2004 (arm, hand), 2007, 2008 (ben), 2013, 2022
Dystrofia myotonika (DM)	2000, 2003, 2007, 2014, 2017, 2023
Ehlers Danlos syndrom (EDS)	2000, 2000, 2001, 2004, 2007, 2010, 2013, 2014, 2015, 2017, 2019, 2022
Ektodermal dysplasi (ED)	2000, 2008, 2015, 2024
Enkammarhjärta	1995, 2013, 2018, 2023
Epidermolysis bullosa (EB)	1995, 2004, 2022
Esofagusatresi	1998, 2005
Fenylketonuri (PKU)	1995, 2002, 2009, 2015, 2021
Fetalt alkoholsyndrom (FAS)	2014
Fragilt X syndrom	1998, 2003, 2006, 2012, 2015, 2019, 2022
Galaktosemi	2004, 2014
Gallvägsatresi	2020
Gauchers sjukdom	2011
Glut-1-brist och PDH-brist	2013
Glutarsyrauri	2017
Goldenhar syndrom	2002
Grav språkstörning	2001, 2001, 2003, 2009
Hemparenteral nutrition	2002
Hereditär spastisk parapares (HSP)	2005
Heterotaxisyndromet	2020
Hjärtsjukdom, skador efter operation	1999
Hjärtrplanterade barn	2000, 2009
Hydrocefalus	2002, 2006
Iktyos	1999, 2003, 2009, 2014, 2019
Immunbristsjukdomar	1998, 2012
Incontinentia pigmenti	2016
Jouberts syndrom	2004, 2012, 2022

Diagnos	Årtal
Kabukisyndromet	2007, 2011, 2022
Kavernom	2023
Klinefelters syndrom	1999, 2003, 2007, 2012, 2019, 2023 (lättläst version)
Kongenital muskeldystrofi	2002, 2009, 2013, 2017
Kongenital binjurebarkshyperplasi	2005, 2010, 2011, 2014, 2018
Kongenital finsk nefros	2006
Kongenitala myopati	2018
Krabbes sjukdom	2013
Kraniofaciala missbildningar	2006, 2007, 2009
Kromosom 18 förändringar	2014
Kromosom 22 förändringar	2016
Kronisk njursjukdom	1996
Landau Kleffners syndrom	1997
Langerhans cellhistiocytos	2005
Laurence-Moon-Bardet-Biedl syndrom (LMBB)	1998, 2005, 2010
LCHAD-brist	2004, 2015
Lennox-Gastauts syndrom	2015
Lesch-Nyhans syndrom	2003
Leukodystrofier	2000, 2014, 2019, 2004, 2014
Leversjukdomar	1997
Limb-girde muskeldystrofi	2007, 2019, 2024
Lissencefali	2018
Loeys-Dietz syndrom	2018
Marfans syndrom	2002, 2007, 2012, 2018
Markörkromosom 15-syndromet	2005, 2016
MECP2-duplikationssyndromet	2017
Medfött diafragmabråck	2023
Metabola sjukdomar	1996
Mitokondriella sjukdomar	1999, 2004, 2007, 2011, 2015, 2018
MPS-sjukdomar	1998, 2003, 2015, 2019
Multipel skleros (MS)	2008
Möbius syndrom	1997, 2004, 2017
Narkolepsi	2012, 2013 april, 2013 maj, 2013, dec, 2014 april, 2014 samlingsdokumentation, 2017, 2020
Nefrotiskt syndrom	2002
Nemalinmyopati	2013, 2021
Neurofibromatos	2000, 2003, 2008, 2012, 2016, 2019, 2022

Diagnos	Årtal
Noonans syndrom	2000, 2005, 2009, 2011, 2013, 2021, 2023 (lättläst version)
Obalanserade translokationer	2018
Tvångssyndrom (OCD)	2003
Optikushypoplasi	2017, 2023
Osteogenesis imperfecta (OI)	2001, 2005, 2010, 2020
PANDAS/PANS	2015
Phelan-McDermids syndrom	2021
PIPO	2024
Pitt Hopkins och Mowat Wilsons syndrom	2014, 2023
Polyneuropatier, ärftliga	1996, 2006
Potocki-Lupskis syndrom	2016, 2021
Prader Willis syndrom	1994, 1998, 2001, 2007, 2011, 2016, 2018, 2022
Retts syndrom	1998, 2000, 2003, 2004, 2006, 2010, 2013, 2016, 2022
Rubinstein-Taybis syndrom	2000, 2004, 2010, 2015, 2021
Ryggmärgsbråck (MMC)	2010, 2013, 2016, 2019
Sallas sjukdom	1997, 2006, 2016
Sanfilippos sjukdom	2010
Septo-optisk dysplasi	1998, 2009
Sicklecellanemi	2022
Silver Russels syndrom	1996, 2002, 2007, 2015, 2019
Sjukdomar i vit hjärnvävnaden	1997
Sjögren-Larssons syndrom	2005
Skelettdysplasier	2015, 2024
Smith-Magenis syndrom	2012, 2022
Sotos syndrom	2000, 2003, 2007, 2016, 2022
Spielmeyer-Vogts sjukdom	1995, 2011, 2014, 2018
Spinal muskelatrofi (SMA)	2001, 2003, 2008, 2015, 2018, 2022, 2023
Stamning	1998
Stroke hos barn och ungdomar	2015, 2019
Sturge-Webers syndrom	2003, 2012, 2019
Svårbehandlad epilepsi	2007, 2008, 2008, 2013, 2014, 2016, 2018, 2020, 2021, 2022
Sällsynta genetiska epilepsier	2020
Tarmsvikt	2017
Tourettes syndrom	1999, 2001, 2002, 2008
Toxoplasmos	1999
Treacher-Collins syndrom	1999, 2005, 2017

Diagnos	Årtal
Tuberös skleros	1999, 2001, 2004, 2011, 2014, 2017, 2020
Turners syndrom	2000, 2003, 2006, 2012, 2016, 2022
Ultrasällsynta syndrom	2023
Uretravalvel	2003
Ushers syndrom	2005, 2019
VACTERL	2001, 2009, 2016
VLCAD-brist	2016
Wiedemann-Steiners syndrom	2023
Williams syndrom	1999, 2002, 2004, 2008, 2012, 2018, 2020, 2023 (lättläst version)