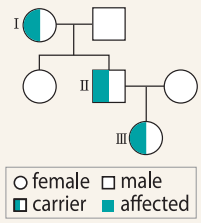
염색체마이크로어레이(CMA) 검사의뢰서

병(의)원명		수진자명		접수번호	
병(의)원코드		생년월일	년 월 일	검체채취일	
진료과/병동		나이/성별		의뢰일	
의사명		차트번호		의뢰기관 연락처	
임상정보(진단명)	(이식력 ☐유 ☐무)			가계도 (가족력)	가계도 [예시]
감염성	☐유 ☐무 (감염성질환명 :)				
인종	☐ East Asian ☐ South Asian ☐ African ☐ Caucasian ☐ Hispanic ☐ Other( ) 혼혈은 중복 표시				
검체종류	☐혈액(EDTA) ☐골수(Bone Marrow) ☐혈액여지 ☐구강점막세포 ☐양수(Amniotic) ☐뇌척수액(CSF) ☐객담(Sputum) ☐소변(Urine) ☐조직(Tissue) ☐기타( )				



임상정보

* 해당되는 임상정보에 표기해 주십시오.

Perinatal Histoy
- ☐ Prematurity
- ☐ Intrauterine growth restriction(IUGR)
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Cystic hygroma
- ☐ Diaphragmatic hernia
- ☐ Omphalocele
- ☐ Encephalocele
- ☐ Neural tube defect
- ☐ Other : _____

Growth
- ☐ Failure to thrive
- ☐ Ovegrowth
- ☐ Short stature
- ☐ Other : _____

Development
- ☐ Fine motor delay
- ☐ Gross motor delay
- ☐ Speech & language delay
- ☐ Developmental regression
- ☐ Pervasive developmental delay
- ☐ Other : _____

Cognitive
- ☐ Intellectual disablility
- ☐ Learning disablility
- ☐ Mental retardation

Behavioral
- ☐ Asperger syndrome features
- ☐ Autism, autistic behavior
- ☐ Autism spectrum disorder
- ☐ Oppositional-defiant disorder (ODD)
- ☐ Obsessive-compulsive disorder (OCD)
- ☐ Attention deficit hyperactivity disorder (ADHD)
- ☐ Other : _____

Neurological
- ☐ Ataxia/dystonia/chorea
- ☐ Hypotonia
- ☐ Anosmia, congenital
- ☐ Seizures
- ☐ Spasticity
- ☐ Cerebral palsy
- ☐ Encelphalopathy
- ☐ Structural brain abnormalities : _____
- ☐ Other : _____

Cardiac
- ☐ Atrial septal defect (ASD)
- ☐ Venticular septal defect (VSD)
- ☐ Atrioventricular (AV) canal defect
- ☐ Coartation of aorta
- ☐ Cardiomyopathy
- ☐ Hypoplastic left heart
- ☐ Tetralogy of fallot

Craniofacial/Dysmorphism
- ☐ Dysmorphic facial features
- ☐ Cleft lip ± cleft palate
- ☐ Cleft palate alone
- ☐ Coloboma
- ☐ Craniosynostosis
- ☐ Ear malformation
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Brachycephaly
- ☐ Other : _____

Cutaneous
- ☐ Hyperpigmentation
- ☐ Hypopigmentation
- ☐ Café-an-lait macules
- ☐ Cutis laxa
- ☐ Ichthyosis
- ☐ Eczema
- ☐ Other : _____

Musculoskeletal
- ☐ Contractures(arthrogryposis)
- ☐ Culb foot
- ☐ Limb anomaly
- ☐ Polydactyly
- ☐ Syndactyly
- ☐ Clinodactyly

- ☐ Talipes equinovarus
- ☐ Vertebral anomaly
- ☐ Hemihypertrophy
- ☐ Pectus excavatum
- ☐ Other : _____

Gastrointestinal
- ☐ Gastroschisis
- ☐ Omphalocele
- ☐ Congenital megacolon
- ☐ Tracheoesophageal fistula
- ☐ Pyloryc stenosis
- ☐ Anal atresia
- ☐ Other : _____

Genitourinary
- ☐ Ambiguous genitalia
- ☐ Hydronephrosis
- ☐ Kidney anomaly : _____
- ☐ Cryptorchidism
- ☐ Urethra/ureter obstruction
- ☐ Hypospadias
- ☐ Other : _____

Family History
- ☐ Parents with 2 or more miscarriages
- ☐ Relatives with known chromosome abnormality : _____
- ☐ Other relatives with similar clinical histoy
- ☐ None

※ 유전자 검사 의뢰시 「유전자 검사 동의서」를 반드시 첨부해야 합니다.

※ 정도관리에서 검사 중 우연히 발견된 소견(Incidental finding)을 통보받으시겠습니까? ☐예 ☐아니오

검사기관기호 : 41341473 | 인천광역시 연수구 하모니로 291 | Tel : 1600-0021 | www.eonelab.co.kr

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이원의료재단 EONE Laboratories

CMA 검사 검출질환의 예

1p36 deletion syndrome (OMIM 607872)
Alagille syndrome (OMIM 118450, 610205)
Congenital ptosis (OMIM 178300)
Deafness (OMIM 608372) (3번 OMIM 606012)
1q21.1 deletion syndrome (OMIM 612474)
1q21.1 duplication syndrome (OMIM 612475)
Split-hand/foot malformation (OMIM 119100)
Pituitary hormone deficiency (OMIM 262700)
van der Woude syndrome (OMIM 119300)
1q43q44 microdeletion syndrome (OMIM 612337)
Basal cell nevus syndrome (OMIM 109400)
Essential tremor (OMIM 602134)
Feingold syndrome (OMIM 164280, 614326)
2p15p16.1 deletion syndrome (OMIM 612513)
Intellectual developmental disorder with autism and speech delay (OMIM 606053)
Mowat-Wilson syndrome (OMIM 235730)
Synpolydactyly (OMIM 186000)
2q31.2 deletion syndrome (OMIM 612345)
2q32q33 deletion syndrome (Glass syndrome) (OMIM 612313)
2q37 deletion syndrome (OMIM 600430)
Developmental dysplasia of the hip (OMIM 615612)
Waardenburg syndrome (OMIM 193510)
Congenital facial paresis (OMIM 601471)
Camptodactyly (OMIM 114200)
Asperger syndrome susceptibility (OMIM 608638)
Blepharophimosis, epicanthus inversus, and ptosis (OMIM 110100)
Dandy-Walker syndrome (OMIM 220200)
3q29 microdeletion syndrome (OMIM 609425)
3q29 microduplication syndrome (OMIM 611936)
Wolf-Hirschhorn syndrome (OMIM 194190)
Facioscapulohumeral muscular dystrophy (OMIM 158900)
Axenfeld-Rieger syndrome (OMIM 180500)
Cri-du-chat syndrome (OMIM 123450)
Cornelia de Lange syndrome (OMIM 122470)
5q 14.3 microdeletion syndrome (OMIM 612881)
Gardner syndrome (OMIM 175100)
Sotos syndrome (OMIM 117550)
Leukodystrophy, adult-onset, autosomal dominant (OMIM 169500)
Lymphatic malformation (OMIM 611944)
Cleidocranial dysplasia (OMIM 119600)
Greig cephalopolysyndactyly syndrome (OMIM 175700)
Saethre-Chotzen syndrome (OMIM 101400)
Williams-Beuren syndrome (OMIM 194050)
Currarino syndrome (OMIM 176450)
CHARGE syndrome (OMIM 214800)
8q21.11 deletion syndrome (OMIM 614230)
Trichorhinophalangeal syndrome (OMIM 190350)
Nablus mask-like facial syndrome (OMIM 608156)
Branchiootorenal syndrome (OMIM 113650)
Langer-Giedion Syndrome (OMIM 190350)
Loeys-Dietz syndrome (OMIM 609192)
Nail-patella syndrome (OMIM 161200)
Tuberous sclerosis (OMIM 191100)
10q22.3q23.2 deletion syndrome (OMIM 612242)
Potocki-Shaffer syndrome (OMIM 601224)
WAGR syndrome (OMIM 194072)
Beckwith-Wiedemann syndrome (OMIM 130650)
Jacobsen syndrome (OMIM 147791)
Buschke-Ollendorff syndrome (OMIM 166700)
Holt-Oram syndrome (OMIM 142900)
Stickler syndrome (OMIM 108300)
Ulnar-mammary syndrome (OMIM 181450)
Retinoblastoma (OMIM 180200)
14q11q22 deletion syndrome (OMIM 613457)
Prader-Willi syndrome (OMIM 176270)
Angelman syndrome (OMIM 105830)
15q13.3 microdeletion syndrome (OMIM 612001)
Marfan syndrome (OMIM 154700)
15q25 deletion syndrome (OMIM 614294)
Polycystic kidney disease (OMIM 600273)
Alpha-thalassemia/mental retardation syndrome (OMIM 141750)
16p11.2 deletion syndrome (OMIM 613444)
Rubinstein-Taybi syndrome (OMIM 180849)
Townes-Brocks syndrome (OMIM 107480)
Miller-Dieker lissencephaly syndrome (OMIM 247200)
Lissencephaly (OMIM 607432)
17p13.1 deletion syndrome (OMIM 613776, 151623)
Smith-Magenis syndrome (OMIM 182290)
Potocki-Lupski syndrome (OMIM 610883)
Charcot-Marie-Tooth disease (OMIM 118220)
Renal cysts and diabetes syndrome (OMIM 137920)
Koolen-De Vries syndrome (OMIM 610443)
Neurofibromatosis (OMIM 162200)
17q23.1-q23.2 deletion syndrome (OMIM 613355)
Campomelic dysplasia (OMIM 114290)
18q deletion syndrome (OMIM 601808)
19p13.13 deletion/duplication syndrome (OMIM 613638)
Cat eye syndrome (OMIM 115470)
22q11.2 microduplication syndrome (OMIM 608363)
22q11.2 deletion syndrome (OMIM 611867)
DiGeorge syndrome/ Velocardiofacial syndrome (OMIM 188400, 192430)
Phelan-McDermid syndrome (OMIM 606232)
Raynaud-Claes syndrome (OMIM 300114)
Alport syndrome (OMIM 301050)
Ichthyosis (OMIM 308100)
Xp11.23p11.22 duplication syndrome (OMIM 300801)
Xq28 duplication syndrome (OMIM 300815)
Rett syndrome (OMIM 312750)
Moyamoya disease 4 (OMIM 300845)
Adrenal hypoplasia, congenital (OMIM 300200)
Glycerol kinase deficiency (OMIM 307030)
Kallmann syndrome (OMIM 308700)

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