

Table 2 Immunological symptoms and comorbidities of 14 patients with 11q-disorders

Patient	IgRT	Viral infections	Bacterial infections	Eczema	Warts	Mycosis	Comorbidities
On Immunoglobulin Replacement Therapy (IgRT)							
1	Yes	Frequent prolonged viral upper respiratory tract infections (URTI) and ear-nose-throat infections (ENTI) at younger age. Varicella-Zoster Virus infection (VZV): caregiver does not recall	Frequent ENTI at young age needing tubes and frequent antibiotics, at age 40 panorychium, needing drainage	None	Many, chronically, on hand and foot. Gardasil needed.	None	Mildly increased bleeding tendency. Lymphangiomyomatosis both lower legs; asymmetric Aortic valve; congenital. Ectropion; hearing aids; mild-moderate developmental delay
2	Since age of 6 yrs	Frequent ENTI and URTI at young age. Frequent rehydration after viral infections and GE-infections. Very severe VZV infections	Frequent AB and tubes for ENTI at young age, panorychium needing drainage, severe bacterial infections of VZV lesions, needing local and iv AB	None	None	Severe fungal nails and vaginal mycosis, chronic use of anti-mucosal treatment	Marked increased bleeding tendency. Moderate-severe developmental delay, craniosynostosis repairment, eyelid surgery. Type 1 diabetes mellitus at age 11 yrs
3	Yes	Recurrent URTI, viral bronchitis and BHR, needing hospitalization for salbutamol and oxygen therapy. VZV at young age, mild-moderate course.	Frequent AB and AB-prophylaxis till age of 6 for frequent ENTI.	None	None	None	Increased bleeding tendency. Large VSD; Eyelid surgery; Nissen-surgery for reflux disease (GERD); moderate-severe developmental delay
7	Since age of 10 yrs	In first 4 years of life frequent hospitalization for dehydration during severe GE-infections. Frequent URTI and ENTI. VZV infection at 2 yrs, moderate course, severe H1N1 influenza 2009, Tamiflu en AB needed.	Frequent AB and tubes for ENTI. Once hospitalization for sepsis * (occurred under IgRT).	None	None	None	Marked increased bleeding tendency. Small gestational age, craniosynostosis repairment surgery, large VSD; severe developmental delay; epilepsy; surgical eye lid correction. Severe GERD. Celiac disease. Sudden death at age of 16 yrs.
11	Since age of 5 yrs	Severe RSV and para-influenza-infection. BHR, needing salbutamol on indication. Frequent hospitalization for viral URTI; dehydration with Rotavirus. Severe VZV at 1 month old.	Bacterial superinfection of the skin during VZV and frequent ENTI needing AB	Mild eczema	None	Pityriasis rosacea, fungal nails needing terbinafine	Marked increased bleeding tendency. Mild left pulmonary artery stenosis; mild developmental delay
14T	Since age of 14 yrs	Frequent URTI and ENTI. Severe VZV infection. Frequent oral aphthous stomatitis	Frequent AB and tubes for ENTI and URTI. Bacterial infections of VZV lesions, needing local AB	Chronic eczema, resolving after start of IgRT	one persistent on toe	Cutaneous ketoconazole for frequent diaper dermatitis	Marked increased bleeding tendency. Moderate-severe developmental delay. Congenital hip dysplasia,

Not on Immunoglobulin Replacement Therapy						
4	No	No prolonged or complicated viral infections	No infections	None	None	None
5	No	No prolonged or complicated viral infections	No infections	None	None	Mildly lower platelet count. Mild increased bleeding tendency. Multiple VSD, feeding difficulties. Mildly delayed development for her age.
6	No	Severe VZV; recurrent URTI but no ENTI; Once GE-it is, needing rehydration	Bacterial superinfection of the skin during VZV.	Frequently skin dermatitis in hair, axillary and umbilical area	None	Skin fungal dermatitis, needing miconazol
8	No	Recurrent ENTI requiring ear tubes. Moderate-severe VZV	On AB prophylaxis since age 4yr for frequent ENTI	Yes eczema	None	Chronically, between toes, needing miconazol
9	No	BHR, needing salbutamol on indication. Once hospitalized for oxygen replacement during viral LRTI. Severe VZV-infection. Frequent ENTI & URTI	Frequent AB and tubes for ENTI. Bacterial superinfection needing AB during VZV	Yes, seborrheic eczema	Chronically in toe-nails	No increased bleeding tendency. Variant HLHS, ADHD, mild cognitive delay
10	No	BHR needing salbutamol on indication, frequent URTI . Moderate VZV at age 18yr	In first 10 years of life; frequent AB for ENTI and prolonged URTI	Frequent seborrheic eczema around ears and in hair	None	Low platelet count, increased bleeding tendency. VSD, pulmonary hypertension, developmental delay, hearing aid needed. Once hospitalization for fever-related convulsion; once for car-accident with mild brain injury, no extra bleeding complications.
12	No	BHR, needing salbutamol on indication, severe pseudocroup at age 6, needing epinephrin. Frequent hospitalizations for URTI (1x picornavirus, 1x rhinovirus) Frequent ENTI and URTI. Not yet VZV	Frequent AB and tubes for ENTI and URTI. Still on AB prophylaxis	Yes	None	Marked increased bleeding tendency. Moderate severe MI, surgical repairment of cleft palate, neonatal apneas and hypotonia, horse shoe kidney, retrognathia, feeding difficulties requiring PEG-tubea, West syndrome, severe developmental and speech delay, osteoporosis, needing biphosphonates iv. Loss of vision after vitreous hemorrhage and retinal ablation
13T	No	Frequent ENTI. No VZV-infection yet.	Sporadic antibiotics. Ear tubes for ENTI and glue ears at age 3yrs. Frequent Urinary infections (UTI),		Frequent diaper dermatitis, needing miconazol	No bleeding tendency. Severe developmental delay, fever-related convulsion at age 6, 9 and 11 years

once complicated by
pyelonephritis

Abbreviations: AB = antibiotics, ADHD = Attention Deficit Hyperactivity Disorder, ASD = Atrial Septum Defect, ATE = Adenotonsillectomy, BHR = Bronchial Hyperreactivity, ENT = Ear-Nose-Throat, F = Female, GERD = Gastro-Esophageal Reflux Disease GI = Gastro-intestinal, IgRT = Immunoglobulin Replacement Therapy, M= Male, HLHS = Hypoplastic Left Heart Syndrome, LRTI = Lower Respiratory Tract Infections, RSV – Respiratory Syncytial Virus, VSD = Ventricular Septum Defect, VZV = Varicella-Zoster Virus, URTI = Upper Respiratory Tract Infections, UTI = urinary tract infections, VSD = ventricular-septum defect,